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

  Email carmitag@stanford.edu

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

2026-27

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

2025-26

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

2023-24

- Informatics in Industry: BIOMEDIN 206 (Spr)

STANFORD ADVISEES

Doctoral Dissertation Reader (AC)

Vidal Arroyo, Jon Bezney, Ziwei Chen, Tania Fabo, Karen Feng, Jodie Lunger, Imani Porter, Jess Rhodes, Alp Tartici

Postdoctoral Faculty Sponsor

Evin Padhi, Yilin Xie

Doctoral Dissertation Advisor (AC)

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

Doctoral Dissertation Co-Advisor (AC)

Jordan Cahoon, Haim Krupkin

Master's Program Advisor

Nate Demchak, Krithika Nathamuni, 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
  2023

- **Author Correction: Africa-specific human genetic variation near CHD1L associates with HIV-1 load.** *Nature*
  McLaren, P. J., Porreca, I., Iaconis, G., Mok, H. P., Mukhopadhyay, S., Karakoc, E., Cristinelli, S., Pomilla, C., Bartha, I., Thorball, C. W., Tough, R. H., Angelino, P., Kiar, et al
  2023

- **Beyond the exome: What's next in diagnostic testing for Mendelian conditions.** *American journal of human genetics*
  Wojcik, M. H., Reuter, C. M., Marwaha, S., Mahmoud, M., Duyzend, M. H., Barseghyan, H., Yuan, B., Boone, P. M., Groopman, E. E., Délot, E. C., Jain, D., Sanchis-Juan, A., Starita, et al
  2023; 110 (8): 1229-1248

- **Africa-specific human genetic variation near CHD1L associates with HIV-1 load.** *Nature*
  McLaren, P. J., Porreca, I., Iaconis, G., Mok, H. P., Mukhopadhyay, S., Karakoc, E., Cristinelli, S., Pomilla, C., Bartha, I., Thorball, C. W., Tough, R. H., Angelino, P., Kiar, et al
  2023

- **Molecular quantitative trait loci** *NATURE REVIEWS METHODS PRIMERS*
  Aguet, F., Alasoo, K., Li, Y., Battle, A., Im, H., Montgomery, S. B., Lappalainen, T.

2023; 3 (1)

- **Beyond the exome: what's next in diagnostic testing for Mendelian conditions.** *ArXiv*
  Wojcik, M. H., Reuter, C. M., Marwaha, S., Mahmoud, M., Duyzend, M. H., Barseghyan, H., Yuan, B., Boone, P. M., Groopman, E. E., Délot, E. C., Jain, D., Sanchis-Juan, A., Starita, et al
  2023

- **The mitochondrial multi-omic response to exercise training across tissues.** *bioRxiv : the preprint server for biology*
  Amar, D., Gay, N. R., Jimenez-Morales, D., Beltran, P. M., Ramaker, M. E., Raja, A. N., Zhao, B., Sun, Y., Marwaha, S., Gaul, D., Hershman, S. G., Xia, A., Lanza, et al
  2023

- **Multiomic identification of key transcriptional regulatory programs during endurance exercise training.** *bioRxiv : the preprint server for biology*
  Smith, G. R., Zhao, B., Lindholm, M. E., Raja, A., Viggars, M., Pincas, H., Gay, N. R., Sun, Y., Ge, Y., Nair, V. D., Sanford, J. A., S Amper, M. A., Vasoya, et al
  2023

- **RNAget: an API to securely retrieve RNA quantifications.** *Bioinformatics (Oxford, England)*
  Upchurch, S., Palumbo, E., Adams, J., Bujold, D., Bourque, G., Nedzel, J., Graham, K., Kagda, M. S., Assis, P., Hitz, B., Righi, E., Guigo, R., Wold, et al
  2023; 39 (4)

- **Methylation differences in Alzheimer's disease neuropathologic change in the aged human brain.** *Acta neuropathologica communications*
  Lang, A. L., Eulalio, T., Fox, E., Yakabi, K., Bukhari, S. A., Kawas, C. H., Corrada, M. M., Montgomery, S. B., Heppner, F. L., Capper, D., Nachun, D., Montine, T. J.
  2022; 10 (1): 174

- **Deep learning-assisted genome-wide characterization of massively parallel reporter assays.** *Nucleic acids research*
  Lu, F., Sossin, A., Abell, N., Montgomery, S. B., He, Z.
  2022

- **RNA editing underlies genetic risk of common inflammatory diseases.** *Nature*
  Li, Q., Gloudemans, M. J., Geisinger, J. M., Fan, B., Aguet, F., Sun, T., Ramaswami, G., Li, Y. I., Ma, J. B., Pritchard, J. K., Montgomery, S. B., Li, J. B.
  2022

- **Temporal dynamics of the multi-omic response to endurance exercise training across tissues**
  Gay, N. R., Beltran, P., Amar, D., Montgomery, S. B., Carr, S. A., Motrpac Study Grp
  ELSEVIER.2022: S31

- **Integration of rare expression outlier-associated variants improves polygenic risk prediction.** *American journal of human genetics*
  Smail, C., Ferraro, N. M., Hui, Q., Durrant, M. G., Aguirre, M., Tanigawa, Y., Keever-Keigher, M. R., Rao, A. S., Justesen, J. M., Li, X., Gloudemans, M. J., Assimes, T. L., Kooperberg, et al
  2022

- **Multiple causal variants underlie genetic associations in humans.** *Science (New York, N.Y.)*
  Abell, N. S., DeGorter, M. K., Gloudemans, M. J., Greenwald, E., Smith, K. S., He, Z., Montgomery, S. B.
  2022; 375 (6586): 1247-1254

- **Integration of genetic colocalizations with physiological and pharmacological perturbations identifies cardiometabolic disease genes.** *Genome medicine*
  Gloudemans, M. J., Balliu, B., Nachun, D., Schnurr, T. M., Durrant, M. G., Ingelsson, E., Wabitsch, M., Quertermous, T., Montgomery, S. B., Knowles, J. W., Carcamo-Orive, I.
  2022; 14 (1): 31

- **Integration of genetic colocalizations with physiological and pharmacological perturbations identifies cardiometabolic disease genes**
  Gloudemans, M. J., Balliu, B., Nachun, D., Durrant, M. G., Ingelsson, E., Wabitsch, M., Quertermous, T., Montgomery, S. B., Knowles, J., Carcamo-Orive, I.
  W B SAUNDERS CO-ELSEVIER INC.2022: S24-S25

- **TOWARDS TRANSCRIPTOMICS AS A PRIMARY TOOL FOR RARE DISEASE INVESTIGATION.** *Cold Spring Harbor molecular case studies*
  Montgomery, S. B., Bernstein, J. A., Wheeler, M. T.
  2022

- **Lymphoid blast transformation in an MPN with BCR-JAK2 treated with ruxolitinib: putative mechanisms of resistance.** *Blood advances*
  Chen, J. A., Hou, Y., Roskin, K. M., Arber, D. A., Bangs, C. D., Baughn, L. B., Cherry, A. M., Ewalt, M. D., Fire, A. Z., Fresard, L., Kearney, H. M., Montgomery, S. B., Ohgami, et al
  2021; 5 (17): 3492-3496

- **Genome-wide functional screen of 3'UTR variants uncovers causal variants for human disease and evolution.** *Cell*
  Griesemer, D., Xue, J. R., Reilly, S. K., Ulirsch, J. C., Kukreja, K., Davis, J. R., Kanai, M., Yang, D. K., Butts, J. C., Guney, M. H., Luban, J., Montgomery, S. B., Finucane, et al
  2021

- **The role of Sp140 revealed in IgE and mast cell responses in Collaborative Cross mice.** *JCI insight*
  Matsushita, K., Li, X., Nakamura, Y., Dong, D., Mukai, K., Tsai, M., Montgomery, S. B., Galli, S. J.
  2021; 6 (12)

- **Identification of putative causal loci in whole-genome sequencing data via knockoff statistics.** *Nature communications*
  He, Z., Liu, L., Wang, C., Le Guen, Y., Lee, J., Gogarten, S., Lu, F., Montgomery, S., Tang, H., Silverman, E. K., Cho, M. H., Greicius, M., Ionita-Laza, et al
  2021; 12 (1): 3152

- **Compound heterozygous KCTD7 variants in progressive myoclonus epilepsy.** *Journal of neurogenetics*
  Burke, E. A., Sturgeon, M., Zastrow, D. B., Fernandez, L., Prybol, C., Marwaha, S., Frothingham, E. P., Ward, P. A., Eng, C. M., Fresard, L., Montgomery, S. B., Enns, G. M., Fisher, et al
  2021: 1–10

- **Population-scale tissue transcriptomics maps long non-coding RNAs to complex disease.** *Cell*
  de Goede, O. M., Nachun, D. C., Ferraro, N. M., Gloudemans, M. J., Rao, A. S., Smail, C., Eulalio, T. Y., Aguet, F., Ng, B., Xu, J., Barbeira, A. N., Castel, S. E., Kim-Hellmuth, et al
  2021

- **Functional and structural analysis of cytokine selective IL6ST defects that cause recessive hyper-IgE syndrome.** *The Journal of allergy and clinical immunology*
  Chen, Y., Zastrow, D. B., Metcalfe, R. D., Gartner, L., Krause, F., Morton, C. J., Marwaha, S., Fresard, L., Huang, Y., Zhao, C., McCormack, C., Bick, D., Worthey, et al
  2021

- **Identification of rare and common regulatory variants in pluripotent cells using population-scale transcriptomics.** *Nature genetics*
  Bonder, M. J., Smail, C., Gloudemans, M. J., Fresard, L., Jakubosky, D., D'Antonio, M., Li, X., Ferraro, N. M., Carcamo-Orive, I., Mirauta, B., Seaton, D. D., Cai, N., Vakili, et al
  2021

- **Evaluating the Genomic Parameters Governing rAAV-Mediated Homologous Recombination** *MOLECULAR THERAPY*
  Spector, L. P., Tiffany, M., Ferraro, N. M., Abell, N. S., Montgomery, S. B., Kay, M. A.
  2021; 29 (3): 1028–46

- **Exploiting the GTEx resources to decipher the mechanisms at GWAS loci.** *Genome biology*
  Barbeira, A. N., Bonazzola, R., Gamazon, E. R., Liang, Y., Park, Y., Kim-Hellmuth, S., Wang, G., Jiang, Z., Zhou, D., Hormozdiari, F., Liu, B., Rao, A., Hamel, et al
  2021; 22 (1): 49

- **Nonsense-mediated decay is highly stable across individuals and tissues.** *American journal of human genetics*
  Teran, N. A., Nachun, D. C., Eulalio, T., Ferraro, N. M., Smail, C., Rivas, M. A., Montgomery, S. B.
  2021

- **An integrated approach to identify environmental modulators of genetic risk factors for complex traits.** *American journal of human genetics*
  Balliu, B., Carcamo-Orive, I., Gloudemans, M. J., Nachun, D. C., Durrant, M. G., Gazal, S., Park, C. Y., Knowles, D. A., Wabitsch, M., Quertermous, T., Knowles, J. W., Montgomery, S. B.
  2021

- **Single-cell epigenomic analyses implicate candidate causal variants at inherited risk loci for Alzheimer's and Parkinson's diseases.** *Nature genetics*
  Corces, M. R., Shcherbina, A., Kundu, S., Gloudemans, M. J., Fresard, L., Granja, J. M., Louie, B. H., Eulalio, T., Shams, S., Bagdatli, S. T., Mumbach, M. R., Liu, B., Montine, et al
  2020
- **The GTEx Consortium atlas of genetic regulatory effects across human tissues** *SCIENCE*
  Aguet, F., Barbeira, A. N., Bonazzola, R., Brown, A., Castel, S. E., Jo, B., Kasela, S., Kim-Hellmuth, S., Liang, Y., Parsana, P., Flynn, E., Fresard, L., Gamazon, et al
  2020; 369 (6509): 1318-+
- **Molecular Transducers of Physical Activity Consortium (MoTrPAC): Mapping the Dynamic Responses to Exercise.** *Cell*
  Sanford, J. A., Nogiec, C. D., Lindholm, M. E., Adkins, J. N., Amar, D., Dasari, S., Drugan, J. K., Fernandez, F. M., Radom-Aizik, S., Schenk, S., Snyder, M. P., Tracy, R. P., Vanderboom, et al
  2020; 181 (7): 1464–74
- **Discovery and quality analysis of a comprehensive set of structural variants and short tandem repeats.** *Nature communications*
  Jakubosky, D., Smith, E. N., D'Antonio, M., Jan Bonder, M., Young Greenwald, W. W., D'Antonio-Chronowska, A., Matsui, H., i2QTL Consortium, Stegle, O., Montgomery, S. B., DeBoever, C., Frazer, K. A., Bonder, M. J., et al
  2020; 11 (1): 2928
- **Properties of structural variants and short tandem repeats associated with gene expression and complex traits.** *Nature communications*
  Jakubosky, D., D'Antonio, M., Bonder, M. J., Smail, C., Donovan, M. K., Young Greenwald, W. W., Matsui, H., i2QTL Consortium, D'Antonio-Chronowska, A., Stegle, O., Smith, E. N., Montgomery, S. B., DeBoever, C., et al
  2020; 11 (1): 2927
- **Transcriptional and Position Effect Contributions to rAAV-Mediated Gene Targeting**
  Spector, L. P., Tiffany, M., Ferraro, N. M., Abell, N. S., Montgomery, S. B., Kay, M. A.
  CELL PRESS.2020: 290
- **Molecular Choreography of Acute Exercise.** *Cell*
  Contrepois, K. n., Wu, S. n., Moneghetti, K. J., Hornburg, D. n., Ahadi, S. n., Tsai, M. S., Metwally, A. A., Wei, E. n., Lee-McMullen, B. n., Quijada, J. V., Chen, S. n., Christle, J. W., Ellenberger, et al
  2020; 181 (5): 1112–30.e16
- **Evaluating the genomic parameters governing rAAV-mediated homologous recombination.** *Molecular therapy : the journal of the American Society of Gene Therapy*
  Spector, L. P., Tiffany, M. n., Ferraro, N. M., Abell, N. S., Montgomery, S. B., Kay, M. A.
  2020
- **The impact of sex on gene expression across human tissues.** *Science (New York, N.Y.)*
  Oliva, M. n., Muñoz-Aguirre, M. n., Kim-Hellmuth, S. n., Wucher, V. n., Gewirtz, A. D., Cotter, D. J., Parsana, P. n., Kasela, S. n., Balliu, B. n., Viñuela, A. n., Castel, S. E., Mohammadi, P. n., Aguet, et al
  2020; 369 (6509)
- **Impact of admixture and ancestry on eQTL analysis and GWAS colocalization in GTEx.** *Genome biology*
  Gay, N. R., Gloudemans, M. n., Antonio, M. L., Abell, N. S., Balliu, B. n., Park, Y. n., Martin, A. R., Musharoff, S. n., Rao, A. S., Aguet, F. n., Barbeira, A. N., Bonazzola, R. n., Hormozdiari, et al
  2020; 21 (1): 233
- **Transcriptomic signatures across human tissues identify functional rare genetic variation.** *Science (New York, N.Y.)*
  Ferraro, N. M., Strober, B. J., Einson, J. n., Abell, N. S., Aguet, F. n., Barbeira, A. N., Brandt, M. n., Bucan, M. n., Castel, S. E., Davis, J. R., Greenwald, E. n., Hess, G. T., Hilliard, et al
  2020; 369 (6509)
- **FAM13A affects body fat distribution and adipocyte function.** *Nature communications*
  Fathzadeh, M. n., Li, J. n., Rao, A. n., Cook, N. n., Chennamsetty, I. n., Seldin, M. n., Zhou, X. n., Sangwung, P. n., Gloudemans, M. J., Keller, M. n., Attie, A. n., Yang, J. n., Wabitsch, et al
  2020; 11 (1): 1465
- **A Bioinformatic Analysis of Integrative Mobile Genetic Elements Highlights Their Role in Bacterial Adaptation.** *Cell host & microbe*

Durrant, M. G., Li, M. M., Siranosian, B. A., Montgomery, S. B., Bhatt, A. S.
2019

- **Genetic regulation of gene expression and splicing during a 10-year period of human aging.** *Genome biology*
  Balliu, B., Durrant, M., Goede, O. d., Abell, N., Li, X., Liu, B., Gloudemans, M. J., Cook, N. L., Smith, K. S., Knowles, D. A., Pala, M., Cucca, F., Schlessinger, et al
  2019; 20 (1): 230

- **COMPREHENSIVE RNA ANALYSIS OF CEREBROSPINAL FLUID FROM LEPTOMENINGEAL METASTASES**
  Polyak, D., Li, Y., Liu, B., Connolly, I., Khoeur, L., Kakusa, B., Johnson, E., Andersen, S., Pan, W., Nagpal, S., Montgomery, S. B., Gephart, M.
  OXFORD UNIV PRESS INC.2019: 62

- **Uganda Genome Resource Enables Insights into Population History and Genomic Discovery in Africa.** *Cell*
  Gurdasani, D., Carstensen, T., Fatumo, S., Chen, G., Franklin, C. S., Prado-Martinez, J., Bouman, H., Abascal, F., Haber, M., Tachmazidou, I., Mathieson, I., Ekoru, K., DeGorter, et al
  2019; 179 (4): 984

- **Atheroprotective roles of smooth muscle cell phenotypic modulation and the TCF21 disease gene as revealed by single-cell analysis.** *Nature medicine*
  Wirka, R. C., Wagh, D., Paik, D. T., Pjanic, M., Nguyen, T., Miller, C. L., Kundu, R., Nagao, M., Coller, J., Koyano, T. K., Fong, R., Woo, Y. J., Liu, et al
  2019

- **Identifying causal variants and genes using functional genomics in specialized cell types and contexts.** *Human genetics*
  Liu, B., Montgomery, S. B.
  2019

- **Disease mechanisms elucidated by genetic regulation of human RPE gene expression**
  Vollrath, D., Liu, B., Calton, M. A., Abell, N. S., Benchorin, G., Gloudemans, M. J., Chen, M., Hu, J., Li, X., Balliu, B., Bok, D., Montgomery, S. B.
  ASSOC RESEARCH VISION OPHTHALMOLOGY INC.2019

- **Genetic analyses of human fetal retinal pigment epithelium gene expression suggest ocular disease mechanisms.** *Communications biology*
  Liu, B., Calton, M. A., Abell, N. S., Benchorin, G., Gloudemans, M. J., Chen, M., Hu, J., Li, X., Balliu, B., Bok, D., Montgomery, S. B., Vollrath, D.
  2019; 2 (1): 186

- **Abundant associations with gene expression complicate GWAS follow-up** *NATURE GENETICS*
  Liu, B., Gloudemans, M. J., Rao, A. S., Ingelsson, E., Montgomery, S. B.
  2019; 51 (5): 768-+

- **Identification of 22 novel loci associated with urinary biomarkers of albumin, sodium, and potassium excretion** *KIDNEY INTERNATIONAL*
  Zanetti, D., Rao, A., Gustafsson, S., Assimes, T. L., Montgomery, S. B., Ingelsson, E.
  2019; 95 (5): 1197–1208

- **Transcriptional and Position Effect Contributions to rAAV-Mediated Gene Targeting**
  Spector, L. P., Tiffany, M., Ferraro, N. M., Abell, N. S., Montgomery, S. B., Kay, M. A.
  CELL PRESS.2019: 294

- **Proficiency Testing of Standardized Samples Shows Very High Interlaboratory Agreement for Clinical Next-Generation Sequencing-Based Oncology Assays** *ARCHIVES OF PATHOLOGY & LABORATORY MEDICINE*
  Merker, J. D., Devereaux, K., Iafrate, A., Kamel-Reid, S., Kim, A. S., Moncur, J. T., Montgomery, S. B., Nagarajan, R., Portier, B. P., Routbort, M. J., Smail, C., Surrey, L. F., Vasalos, et al
  2019; 143 (4): 463–71

- **A toolkit for genetics providers in follow-up of patients with non-diagnostic exome sequencing** *JOURNAL OF GENETIC COUNSELING*
  Zastrow, D. B., Kohler, J. N., Bonner, D., Reuter, C. M., Fernandez, L., Grove, M. E., Fisk, D. G., Yang, Y., Eng, C. M., Ward, P. A., Bick, D., Worthey, E. A., Fisher, et al
  2019; 28 (2): 213–28

- **Identification of 22 novel loci associated withurinary biomarkers of albumin, sodium, andpotassium excretion.** *Kidney international*
  Zanetti, D., Rao, A., Gustafsson, S., Assimes, T. L., Montgomery, S. B., Ingelsson, E.
  2019

- **Abundant associations with gene expression complicate GWAS follow-up.** *Nature genetics*
  Liu, B. n., Gloudemans, M. J., Rao, A. S., Ingelsson, E. n., Montgomery, S. B.
  2019; 51 (5): 768–69

- **SEX DIFFERENCES AT THE MOLECULAR LEVEL: LESSONS FROM THE HUMAN TRANSCRIPTOME**
  Stranger, B., Oliva, M., Gamazon, E., Reverter, F., Wucher, V., Balliu, B., Dumitrascu, B., Parsana, P., Payne, A., Jo, B., Montgomery, S., Battle, A., Ardlie, et al
  ELSEVIER.2019: 1034

- **A toolkit for genetics providers in follow-up of patients with non-diagnostic exome sequencing.** *Journal of genetic counseling*
  Zastrow, D. B., Kohler, J. N., Bonner, D. n., Reuter, C. M., Fernandez, L. n., Grove, M. E., Fisk, D. G., Yang, Y. n., Eng, C. M., Ward, P. A., Bick, D. n., Worthey, E. A., Fisher, et al
  2019; 28 (2): 213–28

- **Genetic analyses of human fetal retinal pigment epithelium gene expression suggest ocular disease mechanisms.** *Communications biology*
  Liu, B., Calton, M. A., Abell, N. S., Benchorin, G., Gloudemans, M. J., Chen, M., Hu, J., Li, X., Balliu, B., Bok, D., Montgomery, S. B., Vollrath, D.
  2019; 2: 186

- **Pathologic gene network rewiring implicates PPP1R3A as a central regulator in pressure overload heart failure.** *Nature communications*
  Cordero, P., Parikh, V. N., Chin, E. T., Erbilgin, A., Gloudemans, M. J., Shang, C., Huang, Y., Chang, A. C., Smith, K. S., Dewey, F., Zaleta, K., Morley, M., Brandimarto, et al
  2019; 10 (1): 2760

- **Identification of rare-disease genes using blood transcriptome sequencing and large control cohorts.** *Nature medicine*
  Frésard, L. n., Smail, C. n., Ferraro, N. M., Teran, N. A., Li, X. n., Smith, K. S., Bonner, D. n., Kernohan, K. D., Marwaha, S. n., Zappala, Z. n., Balliu, B. n., Davis, J. R., Liu, et al
  2019

- **Diagnosing rare diseases after the exome.** *Cold Spring Harbor molecular case studies*
  Fresard, L., Montgomery, S. B.
  2018; 4 (6)

- **Proficiency Testing of Standardized Samples Shows Very High Interlaboratory Agreement for Clinical Next-Generation Sequencing-Based Oncology Assays.** *Archives of pathology & laboratory medicine*
  Merker, J. D., Devereaux, K., Iafrate, A. J., Kamel-Reid, S., Kim, A. S., Moncur, J. T., Montgomery, S. B., Nagarajan, R., Portier, B. P., Routbort, M. J., Smail, C., Surrey, L. F., Vasalos, et al
  2018

- **Genetic Regulatory Mechanisms of Smooth Muscle Cells Map to Coronary Artery Disease Risk Loci** *AMERICAN JOURNAL OF HUMAN GENETICS*
  Liu, B., Pjanic, M., Wang, T., Nguyen, T., Gloudemans, M., Rao, A., Castano, V. G., Nurnberg, S., Rader, D. J., Elwyn, S., Ingelsson, E., Montgomery, S. B., Miller, et al
  2018; 103 (3): 377–88

- **Large-Scale Phenome-Wide Association Study of PCSK9 Variants Demonstrates Protection Against Ischemic Stroke** *CIRCULATION-GENOMIC AND PRECISION MEDICINE*
  Rao, A. S., Lindholm, D., Rivas, M. A., Knowles, J. W., Montgomery, S. B., Ingelsson, E.
  2018; 11 (7): e002162

- **Ubiquitination of ABCE1 by NOT4 in Response to Mitochondrial Damage Links Co-translational Quality Control to PINK1-Directed Mitophagy.** *Cell metabolism*
  Wu, Z., Wang, Y., Lim, J., Liu, B., Li, Y., Vartak, R., Stankiewicz, T., Montgomery, S., Lu, B.
  2018

- **Recurrently Mutated Genes Differ between Leptomeningeal and Solid Lung Cancer Brain Metastases.** *Journal of thoracic oncology : official publication of the International Association for the Study of Lung Cancer*
  Li, Y., Liu, B., Connolly, I. D., Kakusa, B. W., Pan, W., Nagpal, S., Montgomery, S. B., Hayden Gephart, M.
  2018

- **Biallelic Mutations in ATP5F1D, which Encodes a Subunit of ATP Synthase, Cause a Metabolic Disorder** *AMERICAN JOURNAL OF HUMAN GENETICS*

Olahova, M., Yoon, W., Thompson, K., Jangam, S., Fernandez, L., Davidson, J. M., Kyle, J. E., Grove, M. E., Fisk, D. G., Kohler, J. N., Holmes, M., Dries, A. M., Huang, et al
2018; 102 (3): 494–504

- **Genetic Regulatory Mechanisms of Smooth Muscle Cells Map to Coronary Artery Disease Risk Loci.** *American journal of human genetics*
  Liu, B. n., Pjanic, M. n., Wang, T. n., Nguyen, T. n., Gloudemans, M. n., Rao, A. n., Castano, V. G., Nurnberg, S. n., Rader, D. J., Elwyn, S. n., Ingelsson, E. n., Montgomery, S. B., Miller, et al
  2018

- **Functional regulatory mechanism of smooth muscle cell-restricted LMOD1 coronary artery disease locus.** *PLoS genetics*
  Nanda, V. n., Wang, T. n., Pjanic, M. n., Liu, B. n., Nguyen, T. n., Matic, L. P., Hedin, U. n., Koplev, S. n., Ma, L. n., Franzén, O. n., Ruusalepp, A. n., Schadt, E. E., Björkegren, et al
  2018; 14 (11): e1007755

- **Allele-specific expression reveals interactions between genetic variation and environment.** *Nature methods*
  Knowles, D. A., Davis, J. R., Edgington, H., Raj, A., Favé, M., Zhu, X., Potash, J. B., Weissman, M. M., Shi, J., Levinson, D. F., Awadalla, P., Mostafavi, S., Montgomery, et al
  2017

- **Population- and individual- specific regulatory variation in Sardinia** *NATURE GENETICS*
  Pala, M., Zappala, Z., Marongiu, M., Li, X., Davis, J. R., Cusano, R., Crobu, F., Kukurba, K. R., Gloudemans, M. J., Reinier, F., Berutti, R., Piras, M. G., Mulas, et al
  2017; 49 (5): 700-?

- **The impact of structural variation on human gene expression** *NATURE GENETICS*
  Chiang, C., Scott, A. J., Davis, J. R., Tsang, E. K., Li, X., Kim, Y., Hadzic, T., Damani, F. N., Ganel, L., Montgomery, S. B., Battle, A., Conrad, D. F., Hall, et al
  2017; 49 (5): 692-?

- **Overexpression of the Cytokine BAFF and Autoimmunity Risk** *NEW ENGLAND JOURNAL OF MEDICINE*
  Steri, M., Orru, V., Idda, M. L., Pitzalis, M., Pala, M., Zara, I., Sidore, C., Faa, V., Floris, M., Deiana, M., Asunis, I., Porcu, E., Mulas, et al
  2017; 376 (17): 1615-1626

- **PML nuclear bodies contribute to the basal expression of the mTOR inhibitor DDIT4** *SCIENTIFIC REPORTS*
  Salsman, J., Stathakis, A., Parker, E., Chung, D., Anthes, L. E., Koskowich, K. L., Lahsaee, S., Gaston, D., Kukurba, K. R., Smith, K. S., Chute, I. C., Leger, D., Frost, et al
  2017; 7

- **Whole transcriptome sequencing in blood provides a diagnosis of spinal muscular atrophy with progressive myoclonic epilepsy (SMA-PME).** *Human mutation*
  Kernohan, K. D., Frésard, L., Zappala, Z., Hartley, T., Smith, K. S., Wagner, J., Xu, H., McBride, A., Bourque, P. R., Consortium, C. R., Bennett, S. A., Dyment, D. A., Boycott, et al
  2017

- **Small RNA Sequencing in Cells and Exosomes Identifies eQTLs and 14q32 as a Region of Active Export** *G3-GENES GENOMES GENETICS*
  Tsang, E. K., Abell, N. S., Li, X., Anaya, V., Karczewski, K. J., Knowles, D. A., Sierra, R. G., Smith, K. S., Montgomery, S. B.
  2017; 7 (1): 31-39

- **FIRE: functional inference of genetic variants that regulate gene expression.** *Bioinformatics (Oxford, England)*
  Ioannidis, N. M., Davis, J. R., DeGorter, M. K., Larson, N. B., McDonnell, S. K., French, A. J., Battle, A. J., Hastie, T. J., Thibodeau, S. N., Montgomery, S. B., Bustamante, C. D., Sieh, W. n., Whittemore, et al
  2017; 33 (24): 3895–3901

- **Long-read genome sequencing identifies causal structural variation in a Mendelian disease.** *Genetics in medicine : official journal of the American College of Medical Genetics*
  Merker, J. D., Wenger, A. M., Sneddon, T. n., Grove, M. n., Zappala, Z. n., Fresard, L. n., Waggott, D. n., Utiramerur, S. n., Hou, Y. n., Smith, K. S., Montgomery, S. B., Wheeler, M. n., Buchan, et al
  2017

- **Overexpression of the Cytokine BAFF and Autoimmunity Risk.** *New England journal of medicine*
  Steri, M., Orrù, V., Idda, M. L., Pitzalis, M., Pala, M., Zara, I., Sidore, C., Faà, V., Floris, M., Deiana, M., Asunis, I., Porcu, E., Mulas, et al

2017; 376 (17): 1615-1626

- **Cohort-specific imputation of gene expression improves prediction of warfarin dose for African Americans.** *Genome medicine*
  Gottlieb, A. n., Daneshjou, R. n., DeGorter, M. n., Bourgeois, S. n., Svensson, P. J., Wadelius, M. n., Deloukas, P. n., Montgomery, S. B., Altman, R. B.
  2017; 9 (1): 98
- **Incorporation of Biological Knowledge Into the Study of Gene-Environment Interactions.** *American journal of epidemiology*
  Ritchie, M. D., Davis, J. R., Aschard, H. n., Battle, A. n., Conti, D. n., Du, M. n., Eskin, E. n., Fallin, M. D., Hsu, L. n., Kraft, P. n., Moore, J. H., Pierce, B. L., Bien, et al
  2017; 186 (7): 771–77
- **Current Challenges and New Opportunities for Gene-Environment Interaction Studies of Complex Diseases.** *American journal of epidemiology*
  McAllister, K. n., Mechanic, L. E., Amos, C. n., Aschard, H. n., Blair, I. A., Chatterjee, N. n., Conti, D. n., Gauderman, W. J., Hsu, L. n., Hutter, C. M., Jankowska, M. M., Kerr, J. n., Kraft, et al
  2017; 186 (7): 753–61
- **Enhancing GTEx by bridging the gaps between genotype, gene expression, and disease.** *Nature genetics*
  2017; 49 (12): 1664–70
- **The impact of rare variation on gene expression across tissues.** *Nature*
  Li, X. n., Kim, Y. n., Tsang, E. K., Davis, J. R., Damani, F. N., Chiang, C. n., Hess, G. T., Zappala, Z. n., Strober, B. J., Scott, A. J., Li, A. n., Ganna, A. n., Bassik, et al
  2017; 550 (7675): 239–43
- **Genetic effects on gene expression across human tissues.** *Nature*
  Battle, A. n., Brown, C. D., Engelhardt, B. E., Montgomery, S. B.
  2017; 550 (7675): 204–13
- **A TNFRSF14-Fc epsilon RI-mast cell pathway contributes to development of multiple features of asthma pathology in mice** *NATURE COMMUNICATIONS*
  Sibilano, R., Gaudenzio, N., DeGorter, M. K., Reber, L. L., Hernandez, J. D., Starkl, P. M., Zurek, O. W., Tsai, M., Zahner, S., Montgomery, S. B., Roers, A., Kronenberg, M., Yu, et al
  2016; 7
- **Directed evolution using dCas9-targeted somatic hypermutation in mammalian cells.** *Nature methods*
  Hess, G. T., Frésard, L., Han, K., Lee, C. H., Li, A., Cimprich, K. A., Montgomery, S. B., Bassik, M. C.
  2016
- **Small RNA Sequencing in Cells and Exosomes Identifies eQTLs and 14q32 as a Region of Active Export.** *G3 (Bethesda, Md.)*
  Tsang, E. K., Abell, N. S., Li, X., Anaya, V., Karczewski, K. J., Knowles, D. A., Sierra, R. G., Smith, K. S., Montgomery, S. B.
  2016
- **DNA Methylation Profiling of Uniparental Disomy Subjects Provides a Map of Parental Epigenetic Bias in the Human Genome.** *American journal of human genetics*
  Joshi, R. S., Garg, P., Zaitlen, N., Lappalainen, T., Watson, C. T., Azam, N., Ho, D., Li, X., Antonarakis, S. E., Brunner, H. G., Buiting, K., Cheung, S. W., Coffee, et al
  2016; 99 (3): 555-566
- **Impact of the X Chromosome and sex on regulatory variation** *GENOME RESEARCH*
  Kukurba, K. R., Parsana, P., Balliu, B., Smith, K. S., Zappala, Z., Knowles, D. A., Fave, M., Davis, J. R., Li, X., Zhu, X., Potash, J. B., Weissman, M. M., Shi, et al
  2016; 26 (6): 768-777
- **An Efficient Multiple-Testing Adjustment for eQTL Studies that Accounts for Linkage Disequilibrium between Variants** *AMERICAN JOURNAL OF HUMAN GENETICS*
  Davis, J. R., Fresard, L., Knowles, D. A., Pala, M., Bustamante, C. D., Battle, A., Montgomery, S. B.
  2016; 98 (1): 216-224
- **An Efficient Multiple-Testing Adjustment for eQTL Studies that Accounts for Linkage Disequilibrium between Variants.** *American journal of human genetics*

Davis, J. R., Fresard, L., Knowles, D. A., Pala, M., Bustamante, C. D., Battle, A., Montgomery, S. B.
2016; 98 (1): 216-24

- **ORegAnno 3.0: a community-driven resource for curated regulatory annotation.** *Nucleic acids research*
  Lesurf, R., Cotto, K. C., Wang, G., Griffith, M., Kasaian, K., Jones, S. J., Montgomery, S. B., Griffith, O. L.
  2016; 44 (D1): D126-32

- **Integrative functional genomics identifies regulatory mechanisms at coronary artery disease loci.** *Nature communications*
  Miller, C. L., Pjanic, M., Wang, T., Nguyen, T., Cohain, A., Lee, J. D., Perisic, L., Hedin, U., Kundu, R. K., Majmudar, D., Kim, J. B., Wang, O., Betsholtz, et al
  2016; 7: 12092-?

- **Non-Coding Loss-of-Function Variation in Human Genomes** *HUMAN HEREDITY*
  Zappala, Z., Montgomery, S. B.
  2016; 81 (2): 78-87

- **A global reference for human genetic variation** *NATURE*
  Altshuler, D. M., Durbin, R. M., Abecasis, G. R., Bentley, D. R., Chakravarti, A., Clark, A. G., Donnelly, P., Eichler, E. E., Flicek, P., Gabriel, S. B., Gibbs, R. A., Green, E. D., Hurles, et al
  2015; 526 (7571): 68-?

- **The landscape of genomic imprinting across diverse adult human tissues** *GENOME RESEARCH*
  Baran, Y., Subramaniam, M., Biton, A., Tukiainen, T., Tsang, E. K., Rivas, M. A., Pirinen, M., Gutierrez-Arcelus, M., Smith, K. S., Kukurba, K. R., Zhang, R., Eng, C., Torgerson, et al
  2015; 25 (7): 927-936

- **Human genomics. Effect of predicted protein-truncating genetic variants on the human transcriptome.** *Science*
  Rivas, M. A., Pirinen, M., Conrad, D. F., Lek, M., Tsang, E. K., Karczewski, K. J., Maller, J. B., Kukurba, K. R., DeLuca, D. S., Fromer, M., Ferreira, P. G., Smith, K. S., Zhang, et al
  2015; 348 (6235): 666-669

- **Effect of predicted protein-truncating genetic variants on the human transcriptome** *SCIENCE*
  Rivas, M. A., Pirinen, M., Conrad, D. F., Lek, M., Tsang, E. K., Karczewski, K. J., Maller, J. B., Kukurba, K. R., DeLuca, D. S., Fromer, M., Ferreira, P. G., Smith, K. S., Zhang, et al
  2015; 348 (6235): 666-669

- **Genetic conflict reflected in tissue-specific maps of genomic imprinting in human and mouse.** *Nature genetics*
  Babak, T., Deveale, B., Tsang, E. K., Zhou, Y., Li, X., Smith, K. S., Kukurba, K. R., Zhang, R., Li, J. B., van der Kooy, D., Montgomery, S. B., Fraser, H. B.
  2015; 47 (5): 544-549

- **Genetic conflict reflected in tissue-specific maps of genomic imprinting in human and mouse.** *Nature genetics*
  Babak, T., Deveale, B., Tsang, E. K., Zhou, Y., Li, X., Smith, K. S., Kukurba, K. R., Zhang, R., Li, J. B., van der Kooy, D., Montgomery, S. B., Fraser, H. B.
  2015; 47 (5): 544-549

- **RNA Sequencing and Analysis.** *Cold Spring Harbor protocols*
  Kukurba, K. R., Montgomery, S. B.
  2015; 2015 (11): 951-69

- **Tissue-specific effects of genetic and epigenetic variation on gene regulation and splicing.** *PLoS genetics*
  Gutierrez-Arcelus, M., Ongen, H., Lappalainen, T., Montgomery, S. B., Buil, A., Yurovsky, A., Bryois, J., Padioleau, I., Romano, L., Planchon, A., Falconnet, E., Bielser, D., Gagnebin, et al
  2015; 11 (1)

- **RNA Sequencing and Analysis.** *Cold Spring Harbor protocols*
  Kukurba, K. R., Montgomery, S. B.
  2015; 2015 (11): pdb top084970-?

- **Type I interferon signaling genes in recurrent major depression: increased expression detected by whole-blood RNA sequencing.** *Molecular psychiatry*

Mostafavi, S., Battle, A., Zhu, X., Potash, J. B., Weissman, M. M., Shi, J., Beckman, K., Haudenschild, C., McCormick, C., Mei, R., Gameroff, M. J., Gindes, H., Adams, et al
2014; 19 (12): 1267-1274

- **Type I interferon signaling genes in recurrent major depression: increased expression detected by whole-blood RNA sequencing** *MOLECULAR PSYCHIATRY*
  Mostafavi, S., Battle, A., Zhu, X., Potash, J. B., Weissman, M. M., Shi, J., Beckman, K., Haudenschild, C., McCormick, C., Mei, R., Gameroff, M. J., Gindes, H., Adams, et al
  2014; 19 (12): 1267-1274
- **High-Resolution Transcriptome Analysis with Long-Read RNA Sequencing** *PLOS ONE*
  Cho, H., Davis, J., Li, X., Smith, K. S., Battle, A., Montgomery, S. B.
  2014; 9 (9)
- **Transcriptome sequencing of a large human family identifies the impact of rare noncoding variants.** *American journal of human genetics*
  Li, X., Battle, A., Karczewski, K. J., Zappala, Z., Knowles, D. A., Smith, K. S., Kukurba, K. R., Wu, E., Simon, N., Montgomery, S. B.
  2014; 95 (3): 245-256
- **Transcriptome sequencing from diverse human populations reveals differentiated regulatory architecture.** *PLoS genetics*
  Martin, A. R., Costa, H. A., Lappalainen, T., Henn, B. M., Kidd, J. M., Yee, M., Grubert, F., Cann, H. M., Snyder, M., Montgomery, S. B., Bustamante, C. D.
  2014; 10 (8)
- **Transcriptome sequencing from diverse human populations reveals differentiated regulatory architecture.** *PLoS genetics*
  Martin, A. R., Costa, H. A., Lappalainen, T., Henn, B. M., Kidd, J. M., Yee, M., Grubert, F., Cann, H. M., Snyder, M., Montgomery, S. B., Bustamante, C. D.
  2014; 10 (8)
- **Cis and trans effects of human genomic variants on gene expression.** *PLoS genetics*
  Bryois, J., Buil, A., Evans, D. M., Kemp, J. P., Montgomery, S. B., Conrad, D. F., Ho, K. M., Ring, S., Hurles, M., Deloukas, P., Davey Smith, G., Dermitzakis, E. T.
  2014; 10 (7)
- **Cis and trans effects of human genomic variants on gene expression.** *PLoS genetics*
  Bryois, J., Buil, A., Evans, D. M., Kemp, J. P., Montgomery, S. B., Conrad, D. F., Ho, K. M., Ring, S., Hurles, M., Deloukas, P., Davey Smith, G., Dermitzakis, E. T.
  2014; 10 (7): e1004461
- **Determining causality and consequence of expression quantitative trait loci** *HUMAN GENETICS*
  Battle, A., Montgomery, S. B.
  2014; 133 (6): 727-735
- **Allelic Expression of Deleterious Protein-Coding Variants across Human Tissues.** *PLoS genetics*
  Kukurba, K. R., Zhang, R., Li, X., Smith, K. S., Knowles, D. A., How Tan, M., Piskol, R., Lek, M., Snyder, M., MacArthur, D. G., Li, J. B., Montgomery, S. B.
  2014; 10 (5)
- **Dissecting the causal genetic mechanisms of coronary heart disease.** *Current atherosclerosis reports*
  Miller, C. L., Assimes, T. L., Montgomery, S. B., Quertermous, T.
  2014; 16 (5): 406-?
- **SplicePlot: a utility for visualizing splicing quantitative trait loci.** *Bioinformatics*
  Wu, E., Nance, T., Montgomery, S. B.
  2014; 30 (7): 1025-1026
- **Path-scan: a reporting tool for identifying clinically actionable variants.** *Pacific Symposium on Biocomputing. Pacific Symposium on Biocomputing*
  Daneshjou, R., Zappala, Z., Kukurba, K., Boyle, S. M., Ormond, K. E., Klein, T. E., Snyder, M., Bustamante, C. D., Altman, R. B., Montgomery, S. B.
  2014; 19: 229-240
- **Transcriptome analysis reveals differential splicing events in IPF lung tissue.** *PloS one*

Nance, T., Smith, K. S., Anaya, V., Richardson, R., Ho, L., Pala, M., Mostafavi, S., Battle, A., Feghali-Bostwick, C., Rosen, G., Montgomery, S. B.
2014; 9 (5)

- **High-resolution transcriptome analysis with long-read RNA sequencing.** *PloS one*
  Cho, H., Davis, J., Li, X., Smith, K. S., Battle, A., Montgomery, S. B.
  2014; 9 (9)
- **Transcriptome Analysis Reveals Differential Splicing Events in IPF Lung Tissue.** *PloS one*
  Nance, T., Smith, K. S., Anaya, V., Richardson, R., Ho, L., Pala, M., Mostafavi, S., Battle, A., Feghali-Bostwick, C., Rosen, G., Montgomery, S. B.
  2014; 9 (3): e92111
- **Quantifying RNA allelic ratios by microfluidic multiplex PCR and sequencing.** *Nature methods*
  Zhang, R., Li, X., Ramaswami, G., Smith, K. S., Turecki, G., Montgomery, S. B., Li, J. B.
  2014; 11 (1): 51-54
- **Characterizing the genetic basis of transcriptome diversity through RNA-sequencing of 922 individuals** *GENOME RESEARCH*
  Battle, A., Mostafavi, S., Zhu, X., Potash, J. B., Weissman, M. M., McCormick, C., Haudenschild, C. D., Beckman, K. B., Shi, J., Mei, R., Urban, A. E., Montgomery, S. B., Levinson, et al
  2014; 24 (1): 14-24
- **Performance of genomic medicine.** *Genome biology*
  Karczewski, K. J., Montgomery, S. B.
  2013; 14 (12): 316
- **Transcriptome and genome sequencing uncovers functional variation in humans.** *Nature*
  Lappalainen, T., Sammeth, M., Friedländer, M. R., 't Hoen, P. A., Monlong, J., Rivas, M. A., Gonzàlez-Porta, M., Kurbatova, N., Griebel, T., Ferreira, P. G., Barann, M., Wieland, T., Greger, et al
  2013; 501 (7468): 506-511
- **Transcriptome and genome sequencing uncovers functional variation in humans** *NATURE*
  Lappalainen, T., Sammeth, M., Friedlaender, M. R., 't Hoen, P. A., Monlong, J., Rivas, M. A., Gonzalez-Porta, M., Kurbatova, N., Griebel, T., Ferreira, P. G., Barann, M., Wieland, T., Greger, et al
  2013; 501 (7468): 506-511
- **Systematic functional regulatory assessment of disease-associated variants.** *Proceedings of the National Academy of Sciences of the United States of America*
  Karczewski, K. J., Dudley, J. T., Kukurba, K. R., Chen, R., Butte, A. J., Montgomery, S. B., Snyder, M.
  2013; 110 (23): 9607-9612
- **Desktop transcriptome sequencing from archival tissue to identify clinically relevant translocations.** *American journal of surgical pathology*
  Sweeney, R. T., Zhang, B., Zhu, S. X., Varma, S., Smith, K. S., Montgomery, S. B., van de Rijn, M., Zehnder, J., West, R. B.
  2013; 37 (6): 796-803
- **The origin, evolution, and functional impact of short insertion-deletion variants identified in 179 human genomes.** *Genome research*
  Montgomery, S. B., Goode, D. L., Kvikstad, E., Albers, C. A., Zhang, Z. D., Mu, X. J., Ananda, G., Howie, B., Karczewski, K. J., Smith, K. S., Anaya, V., Richardson, R., Davis, et al
  2013; 23 (5): 749-761
- **Examination of the relationship between variation at 17q21 and childhood wheeze phenotypes** *JOURNAL OF ALLERGY AND CLINICAL IMMUNOLOGY*
  Granell, R., Henderson, A. J., Timpson, N., St Pourcain, B., Kemp, J. P., Ring, S. M., Ho, K., Montgomery, S. B., Dermitzakis, E. T., Evans, D. M., Sterne, J. A.
  2013; 131 (3): 685-694
- **Integrating GWAS and Expression Data for Functional Characterization of Disease-Associated SNPs: An Application to Follicular Lymphoma** *AMERICAN JOURNAL OF HUMAN GENETICS*
  Conde, L., Bracci, P. M., Richardson, R., Montgomery, S. B., Skibola, C. F.
  2013; 92 (1): 126-130
- **Passive and active DNA methylation and the interplay with genetic variation in gene regulation.** *eLife*

Gutierrez-Arcelus, M., Lappalainen, T., Montgomery, S. B., Buil, A., Ongen, H., Yurovsky, A., Bryois, J., Giger, T., Romano, L., Planchon, A., Falconnet, E., Bielser, D., Gagnebin, et al
2013; 2

- **Normalizing RNA-Sequencing Data by Modeling Hidden Covariates with Prior Knowledge.** *PloS one*
  Mostafavi, S., Battle, A., Zhu, X., Urban, A. E., Levinson, D., Montgomery, S. B., Koller, D.
  2013; 8 (7)

- **Cancer Transcriptome Sequencing and Analysis** *Cancer Genomics: From Bench to Personalized Medicine*
  Morin, R. D., Montgomery, S. B.
  Elsevier.2013; 1: 31–49

- **Normalizing RNA-sequencing data by modeling hidden covariates with prior knowledge.** *PloS one*
  Mostafavi, S., Battle, A., Zhu, X., Urban, A. E., Levinson, D., Montgomery, S. B., Koller, D.
  2013; 8 (7)

- **Detection and impact of rare regulatory variants in human disease.** *Frontiers in genetics*
  Li, X., Montgomery, S. B.
  2013; 4: 67-?

- **Sex-biased genetic effects on gene regulation in humans** *GENOME RESEARCH*
  Dimas, A. S., Nica, A. C., Montgomery, S. B., Stranger, B. E., Raj, T., Buil, A., Giger, T., Lappalainen, T., Gutierrez-Arcelus, M., McCarthy, M. I., Dermitzakis, E. T.
  2012; 22 (12): 2368-2375

- **Mapping cis- and trans-regulatory effects across multiple tissues in twins** *NATURE GENETICS*
  Grundberg, E., Small, K. S., Hedman, A. K., Nica, A. C., Buil, A., Keildson, S., Bell, J. T., Yang, T., Meduri, E., Barrett, A., Nisbett, J., Sekowska, M., Wilk, et al
  2012; 44 (10): 1084-?

- **Genotype-Based Test in Mapping Cis-Regulatory Variants from Allele-Specific Expression Data** *PLOS ONE*
  Lefebvre, J. F., Vello, E., Ge, B., Montgomery, S. B., Dermitzakis, E. T., Pastinen, T., Labuda, D.
  2012; 7 (6)

- **Patterns of Cis Regulatory Variation in Diverse Human Populations** *PLOS GENETICS*
  Stranger, B. E., Montgomery, S. B., Dimas, A. S., Parts, L., Stegle, O., Ingle, C. E., Sekowska, M., Smith, G. D., Evans, D., Gutierrez-Arcelus, M., Price, A., Raj, T., Nisbett, et al
  2012; 8 (4): 272-284

- **A Systematic Survey of Loss-of-Function Variants in Human Protein-Coding Genes** *SCIENCE*
  MacArthur, D. G., Balasubramanian, S., Frankish, A., Huang, N., Morris, J., Walter, K., Jostins, L., Habegger, L., Pickrell, J. K., Montgomery, S. B., Albers, C. A., Zhang, Z. D., Conrad, et al
  2012; 335 (6070): 823-828

- **Meta-analysis of genome-wide association studies identifies three new risk loci for atopic dermatitis** *NATURE GENETICS*
  Paternoster, L., Standl, M., Chen, C., Ramasamy, A., Bonnelykke, K., Duijts, L., Ferreira, M. A., Alves, A. C., Thyssen, J. P., Albrecht, E., Baurecht, H., Feenstra, B., Sleiman, et al
  2012; 44 (2): 187-192

- **DNA methylation profiles of human active and inactive X chromosomes** *GENOME RESEARCH*
  Sharp, A. J., Stathaki, E., Migliavacca, E., Brahmachary, M., Montgomery, S. B., Dupre, Y., Antonarakis, S. E.
  2011; 21 (10): 1592-1600

- **Epistatic Selection between Coding and Regulatory Variation in Human Evolution and Disease** *AMERICAN JOURNAL OF HUMAN GENETICS*
  Lappalainen, T., Montgomery, S. B., Nica, A. C., Dermitzakis, E. T.
  2011; 89 (3): 459-463

- **Rare and Common Regulatory Variation in Population-Scale Sequenced Human Genomes** *PLOS GENETICS*
  Montgomery, S. B., Lappalainen, T., Gutierrez-Arcelus, M., Dermitzakis, E. T.
  2011; 7 (7)

- **Genome-wide association study identifies a common variant associated with risk of endometrial cancer** *NATURE GENETICS*
  Spurdle, A. B., Thompson, D. J., Ahmed, S., Ferguson, K., Healey, C. S., O'Mara, T., Walker, L. C., Montgomery, S. B., Dermitzakis, E. T., Fahey, P., Montgomery, G. W., Webb, P. M., Fasching, et al
  2011; 43 (5): 451-?

- **From expression QTLs to personalized transcriptomics** *NATURE REVIEWS GENETICS*
  Montgomery, S. B., Dermitzakis, E. T.
  2011; 12 (4): 277-282

- **The Architecture of Gene Regulatory Variation across Multiple Human Tissues: The MuTHER Study** *PLOS GENETICS*
  Nica, A. C., Parts, L., Glass, D., Nisbet, J., Barrett, A., Sekowska, M., Travers, M., Potter, S., Grundberg, E., Small, K., Hedman, A. K., Bataille, V., Bell, et al
  2011; 7 (2)

- **Identification of cis- and trans- regulatory variation modulating microRNA expression levels in human fibroblasts** *GENOME RESEARCH*
  Borel, C., Deutsch, S., Letourneau, A., Migliavacca, E., Montgomery, S. B., Dimas, A. S., Vejnar, C. E., Attar, H., Gagnebin, M., Gehrig, C., Falconnet, E., Dupre, Y., Dermitzakis, et al
  2011; 21 (1): 68-73

- **The functional spectrum of low-frequency coding variation** *GENOME BIOLOGY*
  Marth, G. T., Yu, F., Indap, A. R., Garimella, K., Gravel, S., Leong, W. F., Tyler-Smith, C., Bainbridge, M., Blackwell, T., Zheng-Bradley, X., Chen, Y., Challis, D., Clarke, et al
  2011; 12 (9)

- **A map of human genome variation from population-scale sequencing** *NATURE*
  Altshuler, D., Durbin, R. M., Abecasis, G. R., Bentley, D. R., Chakravarti, A., Clark, A. G., Collins, F. S., De La Vega, F. M., Donnelly, P., Egholm, M., Flicek, P., Gabriel, S. B., Gibbs, et al
  2010; 467 (7319): 1061-1073

- **Genevar: a database and Java application for the analysis and visualization of SNP-gene associations in eQTL studies** *BIOINFORMATICS*
  Yang, T., Beazley, C., Montgomery, S. B., Dimas, A. S., Gutierrez-Arcelus, M., Stranger, B. E., Deloukas, P., Dermitzakis, E. T.
  2010; 26 (19): 2474-2476

- **Integrating common and rare genetic variation in diverse human populations** *NATURE*
  Altshuler, D. M., Gibbs, R. A., Peltonen, L., Dermitzakis, E., Schaffner, S. F., Yu, F., Bonnen, P. E., de Bakker, P. I., Deloukas, P., Gabriel, S. B., Gwilliam, R., Hunt, S., Inouye, et al
  2010; 467 (7311): 52-58

- **Transcriptome genetics using second generation sequencing in a Caucasian population** *NATURE*
  Montgomery, S. B., Sammeth, M., Gutierrez-Arcelus, M., Lach, R. P., Ingle, C., Nisbett, J., Guigo, R., Dermitzakis, E. T.
  2010; 464 (7289): 773-U151

- **Candidate Causal Regulatory Effects by Integration of Expression QTLs with Complex Trait Genetic Associations** *PLOS GENETICS*
  Nica, A. C., Montgomery, S. B., Dimas, A. S., Stranger, B. E., Beazley, C., Barroso, I., Dermitzakis, E. T.
  2010; 6 (4)

- **Out of the sequencer and into the wiki as we face new challenges in genome informatics.** *Genome biology*
  Ning, Z., Montgomery, S. B.
  2010; 11 (10): 308-?

- **Annotating the regulatory genome.** *Methods in molecular biology (Clifton, N.J.)*
  Montgomery, S. B., Kasaian, K., Jones, S. J., Griffith, O. L.
  2010; 674: 313-349

- **The resolution of the genetics of gene expression** *HUMAN MOLECULAR GENETICS*
  Montgomery, S. B., Dermitzakis, E. T.
  2009; 18: R211-R215

- **Common Regulatory Variation Impacts Gene Expression in a Cell Type-Dependent Manner** *SCIENCE*
  Dimas, A. S., Deutsch, S., Stranger, B. E., Montgomery, S. B., Borel, C., Attar-Cohen, H., Ingle, C., Beazley, C., Arcelus, M. G., Sekowska, M., Gagnebin, M., Nisbett, J., Deloukas, et al

2009; 325 (5945): 1246-1250

- **Is the thrifty genotype hypothesis supported by evidence based on confirmed type 2 diabetes- and obesity-susceptibility variants?** *DIABETOLOGIA*
  Southam, L., Soranzo, N., Montgomery, S. B., Frayling, T. M., McCarthy, M. I., Barroso, I., Zeggini, E.
  2009; 52 (9): 1846-1851

- **Current computational methods for prioritizing candidate regulatory polymorphisms.** *Methods in molecular biology (Clifton, N.J.)*
  Montgomery, S.
  2009; 569: 89-114

- **ORegAnno: an open-access community-driven resource for regulatory annotation** *NUCLEIC ACIDS RESEARCH*
  Griffith, O. L., Montgomery, S. B., Bernier, B., Chu, B., Kasaian, K., Aerts, S., Mahony, S., Sleumer, M. C., Bilenky, M., Haeussler, M., Griffith, M., Gallo, S. M., Giardine, et al
  2008; 36: D107-D113

- **Text-mining assisted regulatory annotation** *GENOME BIOLOGY*
  Aerts, S., Haeussler, M., Van Vooren, S., Griffith, O. L., Hulpiau, P., Jones, S. J., Montgomery, S. B., Bergman, C. M.
  2008; 9 (2)

- **Population genomics of human gene expression** *NATURE GENETICS*
  Stranger, B. E., Nica, A. C., Forrest, M. S., Dimas, A., Bird, C. P., Beazley, C., Ingle, C. E., Dunning, M., Flicek, P., Koller, D., Montgomery, S., Tavare, S., Deloukas, et al
  2007; 39 (10): 1217-1224

- **A survey of genomic properties for the detection of regulatory polymorphisms** *PLOS COMPUTATIONAL BIOLOGY*
  Montgomery, S. B., Griffith, O. L., Schuetz, J. M., Brooks-Wilson, A., Jones, S. J.
  2007; 3 (6): 1000-1010

- **ORegAnno: an open access database and curation system for literature-derived promoters, transcription factor binding sites and regulatory variation** *BIOINFORMATICS*
  Montgomery, S. B., Griffith, O. L., Sleumer, M. C., Bergman, C. M., Bilenky, M., Pleasance, E. D., Prychyna, Y., Zhang, X., Jones, S. J.
  2006; 22 (5): 637-640

- **cisRED: a database system for genome-scale computational discovery of regulatory elements** *NUCLEIC ACIDS RESEARCH*
  Robertson, G., Bilenky, M., Lin, K., He, A., Yuen, W., Dagpinar, M., Varhol, R., Teague, K., Griffith, O. L., Zhang, X., Pan, Y., Hassel, M., Sleumer, et al
  2006; 34: D68-D73

- **An application of peer-to-peer technology to the discovery, use and assessment of bioinformatics programs** *NATURE METHODS*
  Montgomery, S. B., Fu, T., Guan, J., Lin, K., Jones, S. J.
  2005; 2 (8): 563-563

- **Sockeye: A 3D environment for comparative genomics** *GENOME RESEARCH*
  Montgomery, S. B., Astakhova, T., Bilenky, M., Birney, E., Fu, T., Hassel, M., Melsopp, C., Rak, M., Robertson, A. G., Sleumer, M., Siddiqui, A. S., Jones, S. J.
  2004; 14 (5): 956-962

- **The genome sequence of the SARS-associated coronavirus** *SCIENCE*
  Marra, M. A., Jones, S. J., Astell, C. R., Holt, R. A., Brooks-Wilson, A., Butterfield, Y. S., Khattra, J., Asano, J. K., Barber, S. A., Chan, S. Y., Cloutier, A., Coughlin, S. M., Freeman, et al
  2003; 300 (5624): 1399-1404