Stanford

Stephen B. Montgomery

Associate Professor of Pathology, and of Genetics

CONTACT INFORMATION

- **Alternate Contact**

  Cevan Smith - Administrative Assistant

  Email csmitty@stanford.edu

Bio

ACADEMIC APPOINTMENTS

- Associate Professor, Pathology
- Associate Professor, Genetics
- Member, Bio-X
- Member, Cardiovascular Institute
- Member, Maternal & Child Health Research Institute (MCHRI)

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/

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

2017-18

- Next Generation Sequencing and Applications: BIOS 201 (Win)

2016-17

- Genetics and Developmental Biology Training Camp: DBIO 200, GENE 200 (Aut)
- Next Generation Sequencing and Applications: BIOS 201 (Win)

STANFORD ADVISEES

Doctoral Dissertation Reader (AC)

Rachel Agoglia, Daniel Cotter, Laura Donohue, John Gorzynski, Robin Meyers, Stephanie Nevins, Roshni Patel, Ben Siranosian, Siming Zhang

Doctoral Dissertation Advisor (AC)

Nathan Abell, Tiffany Eulalio, Nicole Ferraro, Nicole Gay, Mike Gloudemans, Page Goddard, Emily Greenwald, Abhiram Rao, Craig Smail, Nikki Teran, Rachel Ungar

Doctoral Dissertation Co-Advisor (AC)

Matt Durrant, Kameron Rodrigues, Olivia de Goede

Master's Program Advisor

Luis Govea Moreno, Anirudh Joshi, Derek Jow, Makena Low, Gauri Prasad, Cooper Raterink

Postdoctoral Research Mentor

Daniel Nachun

GRADUATE AND FELLOWSHIP PROGRAM AFFILIATIONS

- Biomedical Informatics (Phd Program)
- Genetics (Phd Program)

Publications

PUBLICATIONS

- **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., Wu, S., Moneghetti, K. J., Hornburg, D., Ahadi, S., Tsai, M. S., Metwally, A. A., Wei, E., Lee-McMullen, B., Quijada, J. V., Chen, S., Christle, J. W., Ellenberger, et al
  2020; 181 (5): 1112–30.e16

- **FAM13A affects body fat distribution and adipocyte function.** *Nature communications*
  Fathzadeh, M., Li, J., Rao, A., Cook, N., Chennamsetty, I., Seldin, M., Zhou, X., Sangwung, P., Gloudemans, M. J., Keller, M., Attie, A., Yang, J., 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

- **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., Gloudemans, M. J., Rao, A. S., Ingelsson, E., Montgomery, S. B.
  2019; 51 (5): 768–69

- **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

- **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., 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

- **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., Smail, C., Ferraro, N. M., Teran, N. A., Li, X., Smith, K. S., Bonner, D., Kernohan, K. D., Marwaha, S., Zappala, Z., Balliu, B., 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

- **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., 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

- **Functional regulatory mechanism of smooth muscle cell-restricted LMOD1 coronary artery disease locus.** *PLoS genetics*
  Nanda, V., Wang, T., Pjanic, M., Liu, B., Nguyen, T., Matic, L. P., Hedin, U., Koplev, S., Ma, L., Franzén, O., Ruusalepp, A., 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., 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., Grove, M., Zappala, Z., Fresard, L., Waggott, D., Utiramerur, S., Hou, Y., Smith, K. S., Montgomery, S. B., Wheeler, M., 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., Daneshjou, R., DeGorter, M., Bourgeois, S., Svensson, P. J., Wadelius, M., Deloukas, P., 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., Battle, A., Conti, D., Du, M., Eskin, E., Fallin, M. D., Hsu, L., Kraft, P., 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., Mechanic, L. E., Amos, C., Aschard, H., Blair, I. A., Chatterjee, N., Conti, D., Gauderman, W. J., Hsu, L., Hutter, C. M., Jankowska, M. M., Kerr, J., 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., Kim, Y., Tsang, E. K., Davis, J. R., Damani, F. N., Chiang, C., Hess, G. T., Zappala, Z., Strober, B. J., Scott, A. J., Li, A., Ganna, A., Bassik, et al
  2017; 550 (7675): 239–43

- **Genetic effects on gene expression across human tissues.** *Nature*
  Battle, A., 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

- **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-?

- **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

- **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

- **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

- **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)

- **Performance of genomic medicine.** *Genome biology*
  Karczewski, K. J., Montgomery, S. B.
  2013; 14 (12): 316

- **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

- **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