STEPHEN B. MONTGOMERY *last updated 08/28/2020*

EDUCATION

Doctor of Philosophy, Genetics Sep/2002 – Oct/2006
University of British Columbia, Vancouver, Canada

Bachelor of Applied Science, Engineering Physics Sep/1997 – Apr/2002
Electrical Engineering Option
University of British Columbia, Vancouver, Canada

PROFESSIONAL EXPERIENCE

Associate Professor Jul/2018 – present
Stanford University School of Medicine, Stanford, California, USA
Department of Pathology
Department of Genetics
Department of Computer Science, by courtesy
Director of Genome Informatics, Department of Pathology

Assistant Professor Jul/2011 – Jun/2018
Stanford University School of Medicine, Stanford, California, USA
Department of Pathology, Genetics, Computer Science, by courtesy
Director of Genome Informatics, Department of Pathology

Postdoctoral Researcher Nov/2009 – Jun/2011
University of Geneva, Geneva, Switzerland
Department of Genetic Medicine and Development
Prof. Emmanouil T. Dermitzakis Laboratory

Postdoctoral Researcher Nov/2006 – Oct/2009
Wellcome Trust Sanger Institute, Cambridge, United Kingdom
Human Genetics programme
Prof. Emmanouil T. Dermitzakis Laboratory

Graduate Student Sep/2002 – Oct/2006
Canada's Michael Smith Genome Sciences Centre, Vancouver, Canada
Bioinformatics group
Prof. Steven J.M. Jones Laboratory

Instructor Sep/2002 – Oct/2006
Canadian Bioinformatics Workshops
Bioinformatics, Genomics and Developing the Tools

Undergraduate Assistant Oct/2001 – Apr/2002
PATSCAN – Patent Search Office
UBC Library Services, Vancouver, Canada

Undergraduate Assistant Sep/2001 – Apr/2002
UBC Department of Forestry, Vancouver, Canada

Engineering Intern May/2001– Sep/2001
Department of National Defense, Ottawa, Canada
UBC Engineering Co-op Work Experience

Engineering Intern Sep/2000 – Apr/2001
User Interface Session Management
Raytheon Systems Canada Ltd., Richmond, Canada
UBC Engineering Co-op Work Experience

Engineering Intern May/2000–Aug/2000
Machines and Control Systems
Laser Zentrum Hanover e.V., Hanover, Germany
UBC Engineering Co-op Work Experience

Engineering Intern May/1999 – Apr/2000
Systems Integration and Test
Raytheon Systems Canada Ltd., Richmond, Canada
UBC Engineering Co-op Work Experience

Engineering Intern Jan/1999 – Apr/1999
Software Configuration Management
Raytheon Systems Canada Ltd., Richmond, Canada
UBC Engineering Co-op Work Experience

Publication list also available at:
https://scholar.google.com/citations?user=117h3CAAAAAJ&hl=en

PREPRINT PUBLICATIONS UNDER REVIEW*

***Underlined highlights myself or people in my laboratory.**

1. Marc Jan Bonder[1], Craig Smail[1], Michael J. Gloudemans, Laure Frésard, David Jakubosky, Matteo D'Antonio, Xin Li, Nicole M. Ferraro, Ivan Carcamo-Orive, Bogdan Mirauta, Daniel D. Seaton, Na Cai, Danilo Horta, HipSci Consortium, iPSCORE Consortium, GENESiPS Consortium, PhLiPS Consortium, Erin N. Smith, Kelly A. Frazer, **Stephen B. Montgomery***, Oliver Stegle* Systematic assessment of regulatory effects of human disease variants in pluripotent cells [1]joint first authors [*]joint corresponding authors

2. O. M. de Goede*, N. M. Ferraro, D. C. Nachun, A. S. Rao, F. Aguet, A. N. Barbeira, S. E. Castel, S. Kim-Hellmuth, Y. Park, A. J. Scott, B. J. Strober, GTEx Consortium, C. D. Brown, X. Wen, I. M. Hall, A. Battle, T. Lappalainen, H. K. Im, K. G. Ardlie, T. Quertermous, K. Kirkegaard, **S. B. Montgomery*** Long non-coding RNA gene regulation and trait associations across human tissues [*]joint corresponding authors

PUBLICATIONS UNDER REVIEW*

***Underlined highlights myself or people in my laboratory.**

1. co-author. Africa-specific human genetic variation in CHD1L regulates HIV-1, *in revision*

2. co-author. Transcriptional and Position Effect Contributions to rAAV-Mediated Gene Targeting, *in revision*

PEER-REVIEWED PUBLICATIONS*

***Underlined highlights myself or people in my laboratory.**

1. M Ryan Corces, Anna Shcherbina, Soumya Kundu, Michael J Gloudemans, Laure Frésard, Jeffrey M Granja, Bryan H Louie, Shadi Shams, S Tansu Bagdatli, Maxwell R Mumbach, Bosh Liu, Kathleen S Montine, William J Greenleaf, Anshul Kundaje, **Stephen B Montgomery,** Howard Y Chang, Thomas J Montine (2020), Single-cell epigenomic identification of inherited risk loci in Alzheimer's and Parkinson's disease. Nature Genetics, *in press* Role : Aided analysis and manuscript review.

2. Meritxell Oliva[1], Manuel Muñoz-Aguirre[1], Sarah Kim-Hellmuth[1], Valentin Wucher, Ariel D. H. Gewirtz, Daniel J. Cotter, Princy Parsana, Silva Kasela, Brunilda Balliu, Ana Viñuela, Stephane E. Castel, Pejman Mohammadi, François Aguet, Yuxin Zou, Ekaterina A. Khramtsova, Andrew D. Skol, Diego Garrido-Martín, Ferran Reverter, Andrew Brown, Patrick Evans, Eric R. Gamazon, Anthony Payne, Rodrigo Bonazzola, Alvaro N. Barbeira, Andrew R. Hamel, Angel Martinez-Perez, José Manuel Soria, GTEx Consortium, Brandon L. Pierce, Matthew Stephens, Eleazar Eskin, Emmanouil T. Dermitzakis, Ayellet V. Segrè, Hae Kyung Im, Barbara E. Engelhardt, Kristin G. Ardlie, **Stephen B. Montgomery**, Alexis J. Battle, Tuuli Lappalainen, Roderic Guigó, Barbara E. Stranger. (2020) The impact of sex on gene expression across human tissues. Science, *in press*

3. Nicole M. Ferraro[1], Benjamin J. Strober[1], Jonah Einson, Xin Li, Francois Aguet, Alvaro N. Barbeira, Stephane E. Castel, Joe R. Davis, Austin T. Hilliard, Bence Kotis, YoSon Park, Alexandra J. Scott, Craig Smail, Emily K. Tsang, Kristin G. Ardlie, Themistocles L. Assimes, Ira Hall, Hae Kyung Im, GTEx Consortium, Tuuli Lappalainen, Pejman Mohammadi*, **Stephen B. Montgomery***, Alexis Battle* (2020) Diverse transcriptomic signatures across human tissues identify functional rare genetic variation [1]joint first authors [*]joint corresponding authors, Science*, in press*

4. François Aguet[1, *], Alvaro N Barbeira[1], Rodrigo Bonazzola[1], Andrew Brown[1], Stephane E Castel[1], Brian Jo[1], Silva Kasela[1], Sarah Kim-Hellmuth[1], Yanyu Liang[1], Meritxell Oliva[1], Princy E Parsana[1], Elise Flynn, Laure Fresard, Eric R Gaamzon, Andrew R Hamel, Yuan He, Farhad Hormozdiari, Pejman Mohammadi, Manuel Muñoz-Aguirre, YoSon Park, Ashis Saha, Ayellet V Segrć, Benjamin J Strober, Xiaoquan Wen, Valentin Wucher, Sayantan Das, Diego Garrido-Martín, Nicole R Gay, Robert E Handsaker, Paul J Hoffman, Seva Kashin, Alan Kwong, Xiao Li, Daniel MacArthur, John M Rouhana, Matthew Stephens, Ellen Todres, Ana Viñuela, Gao Wang, Yuxin Zou, The GTEx Consortium, Christopher D Brown, Nancy Cox, Emmanouil Dermitzakis, Barbara E Engelhardt, Gad Getz, Roderic Guigo, **Stephen B Montgomery**, Barbara E Stranger, Hae Kyung Im, Alexis Battle, Kristin G Ardlie*, Tuuli Lappalainen* (2020) The GTEx Consortium atlas of genetic regulatory effects across human tissues [1]joint first authors [*]joint corresponding authors, Science, *in press*

5. Nicole R. Gay, Michael Gloudemans, Margaret L. Antonio, Nathan S. Abell, Brunilda Balliu, YoSon Park, Alicia R. Martin, Shaila Musharoff, Abhiram Rao, François Aguet, Alvaro Barbeira, Rodrigo Bonazzola, Farhad Hormozdiari, GTEx Consortium, Kristin G. Ardlie, Christopher D. Brown, Hae Kyung Im, Tuuli Lappalainen, Xiaoquan Wen, **Stephen B. Montgomery** (2020) Impact of admixture and ancestry on eQTL analysis and GWAS colocalization in GTEx. Genome Biology, *in press*

6. Sanford JA, Nogiec CD, Lindholm ME, Adkins JN, Amar D, Dasari S, Drugan JK, Fernández FM, Radom-Aizik S, Schenk S, Snyder MP, Tracy RP, Vanderboom P, Trappe S, Walsh MJ; Molecular Transducers of Physical Activity Consortium. (2020) Molecular Transducers of Physical Activity Consortium (MoTrPAC): Mapping the Dynamic Responses to Exercise. Cell. 2020 Jun 25;181(7):1464-1474. Role : Study design, manuscript review, co-PI of MoTrPAC site.

7. Jakubosky D, Smith EN, D'Antonio M, Jan Bonder M, Young Greenwald WW, D'Antonio-Chronowska A, Matsui H; i2QTL Consortium, Stegle O, Montgomery SB, DeBoever C, Frazer KA. (2020) Discovery and quality analysis of a comprehensive set of structural variants and short tandem repeats. Nat Commun. 2020 Jun 10;11(1):2928 Role : Aided in analysis and manuscript review.

8. Jakubosky D, D'Antonio M, Bonder MJ, Smail C, Donovan MKR, Young Greenwald WW, Matsui H; i2QTL Consortium, D'Antonio-Chronowska A, Stegle O, Smith EN, **Montgomery SB**, DeBoever C, Frazer KA. (2020) Properties of structural variants and short tandem repeats associated with gene expression and complex traits. Nat Commun. 2020 Jun 10;11(1):2927. Role : Aided in analysis and manuscript review.

9. Contrepois K, Wu S, Moneghetti KJ, Hornburg D, Ahadi S, Tsai MS, Metwally AA, Wei E, Lee-McMullen B, Quijada JV, Chen S, Christle JW, Ellenberger M, Balliu B, Taylor S, Durrant MG, Knowles DA, Choudhry H, Ashland M, Bahmani A, Enslen B, Amsallem M, Kobayashi Y, Avina M, Perelman D, Schüssler-Fiorenza Rose SM, Zhou W, Ashley EA, **Montgomery SB,** Chaib H, Haddad F, Snyder MP (2020) Molecular Choreograhy of Acute Exercise. Cell. 2020 May 28;181(5):1112-1130.e16. Role : Aided in analysis (dASE method) and manuscript review.

10. Fathzadeh M[1], Li J[1], Rao A[1], Cook N, Chennamsetty I, Seldin M, Zhou X, Sangwung P, Gloudemans MJ, Keller M, Attie A, Yang J, Wabitsch M, Carcamo-Orive I, Tada Y, Lusis AJ, Shin MK, Molony CM, McLaughlin T, Reaven G, **Montgomery SB**, Reilly D, Quertermous T, Ingelsson E[*], Knowles JW[*]. (2020) FAM13A affects body fat distribution and adipocyte function. Nat Commun. 2020 Mar 19;11(1):1465. Role : Aided in analysis and manuscript review. [1]joint first authors [*]joint corresponding authors

11. Durrant MG, Li MM, Siranosian BA, **Montgomery SB,** Bhatt AS. (2020) A Bioinformatic Analysis of Integrative Mobile Genetic Elements Highlights Their Role in Bacterial Adaptation, Cell Host Microbe. 2020 Jan 8;27(1):140-153.e9 Role : Aided in analysis and manuscript review.

12. Brunilda Balliu, Matthew Durrant, Olivia de Goede, Nathan Abell, Xin Li, Boxiang Liu, Michael J. Gloudemans, Naomi L. Cook, Kevin S. Smith, Mauro Pala, Francesco Cucca, David Schlessinger, Siddhartha Jaiswal, Chiara Sabatti, Lars Lind, Erik Ingelsson[*], **Stephen B Montgomery**[*] (2019) Genetic dysregulation of gene expression and splicing during a ten-year period of human aging, *Genome Biology*, Nov 4;20(1):230 [*]joint corresponding authors

13. Gurdasani D, Carstensen T, Fatumo S, Chen G, Franklin CS, Prado-Martinez J, Bouman H, Abascal F, Haber M, Tachmazidou I, Mathieson I, Ekoru K, DeGorter MK, Nsubuga RN, Finan C, Wheeler E, Chen L, Cooper DN, Schiffels S, Chen Y, Ritchie GRS, Pollard MO, Fortune MD, Mentzer AJ, Garrison E, Bergström A, Hatzikotoulas K, Adeyemo A, Doumatey A, Elding H, Wain LV, Ehret G, Auer PL, Kooperberg CL, Reiner AP, Franceschini N, Maher D, **Montgomery SB,** Kadie C, Widmer C, Xue Y, Seeley J, Asiki G, Kamali A, Young EH, Pomilla C, Soranzo N, Zeggini E, Pirie F, Morris AP, Heckerman D, Tyler-Smith C, Motala AA, Rotimi C, Kaleebu P, Barroso I, Sandhu MS. (2019) Uganda Genome Resource Enables Insights into Population History and Genomic Discovery in Africa, Cell, 2019. Oct 31;179(4):984-1002.e36. Rôle : Aided in analysis and manuscript review.

14. Wirka RC, Wagh D, Paik DT, Pjanic M, Nguyen T, Miller CL, Kundu R, Nagao M, Coller J, Koyano TK, Fong R, Woo YJ, Liu B, **Montgomery SB**, Wu JC, Zhu K, Chang R, Alamprese M, Tallquist MD, Kim JB, Quertermous T. Atheroprotective roles of smooth muscle cell phenotypic modulation and the TCF21 disease gene as revealed by single-cell analysis. Nat Med. 2019 Aug;25(8):1280-1289. doi: 10.1038/s41591-019-0512-5. Role: Aided in analyses and manuscript review.

15. Cordero P, Parikh VN, Chin ET, Erbilgin A, Gloudemans MJ, Shang C, Huang Y, Chang AC, Smith KS, Dewey F, Zaleta K, Morley M, Brandimarto J, Glazer N, Waggott D, Pavlovic A, Zhao M, Moravec CS, Tang WHW, Skreen J, Malloy C, Hannenhalli S, Li H, Ritter S, Li M, Bernstein D, Connolly A, Hakonarson H, Lusis AJ, Margulies KB, Depaoli-Roach AA, **Montgomery SB**, Wheeler MT, Cappola T, Ashley EA. (2019) Pathologic gene network rewiring implicates PPP1R3A as a central regulator in pressure overload heart failure. Nat Commun. 2019 Jun 24;10(1):2760. doi: 10.1038/s41467-019-10591-5 Role: Aided in data production, analysis and manuscript review.

16. Laure Fresard, Craig Smail, Kevin S. Smith, Nicole M. Ferraro, Nicole A. Teran, Kristin D. Kernohan, Devon Bonner, Xin Li, Shruti Marwaha, Zachary Zappala, Brunilda Balliu, Joe R. Davis, Boxiang Liu, Cameron J. Prybol, Jennefer N. Kholer, Diane B. Zastrow, Dianna G. Fisk, Megan E. Grove, Jean M. Davidson, Taila Hartley, Ruchi Joshi, Benjamin J. Strober, Sowmithri Utiramerur, Care4Rare Canada Consortium, Undiagnosed Diseases Network, Lars Lind, Erik Ingelsson, Alexis Battle, Gill Bejerano, Jonathan A. Bernstein, Euan A. Ashley, Kym M. Boycott, Jason D. Merker, Matthew T. Wheeler, **Stephen B. Montgomery** (2019) Identification of rare-disease genes in diverse undiagnosed cases using whole blood transcriptome sequencing and large control cohorts, Nature Medicine, 2019 Jun;25(6):911-919. doi: 10.1038/s41591-019-0457-8

17. Liu, B.*,[1], Gloudemans, M.[1], Rao, A., Ingelsson, E., **Montgomery, S.B.*** (2019) Abundant associations with gene expression complicate GWAS follow-up. Nature Genetics, May;51(5):768-769. [1]joint first authors *joint corresponding authors

18. Merker JD, Devereaux K, Iafrate AJ, Kamel-Reid S, Kim AS, Moncur JT, **Montgomery SB**, Nagarajan R, Portier BP, Routbort MJ, Smail C, Surrey LF, Vasalos P, Lazar AJ, Lindeman NI. (2019) Proficiency Testing of Standardized Samples Shows Very High Interlaboratory Agreement for Clinical Next-Generation Sequencing-Based Oncology Assays. Arch Pathol Lab Med. 2019 Apr;143(4):463-471. doi: 10.5858/arpa.2018-0336-CP. Role: Aided in analyses and manuscript review.

19. Boxiang Liu, Melissa A. Calton, Nathan S. Abell, Gillie Benchorin, Michael J. Gloudemans, Ming Chen, Jane Hu, Xin Li, Brunilda Balliu, Dean Bok, **Stephen B. Montgomery***, Douglas Vollrath* (2018) Genetic analyses of human fetal retinal pigment epithelium gene expression suggest ocular disease mechanisms**.** Commun Biol. 2019 May 20;2:186. doi: 10.1038/s42003-019-0430-6. *joint corresponding authors

20. Zanetti, D., Rao, A., Gustafsson, S., Assimes, T., **Montgomery, S.B.,** Ingelsson, E. (2019) Identification of 22 novel loci associated with urinary biomarkers of albumin, sodium, and potassium excretion. (on bioRxiv), Kidney Int. 2019 May;95(5):1197-1208. doi: 10.1016/j.kint.2018.12.017. Role: Aided in analyses and manuscript review.

21. Nanda V, Wang T, Pjanic M, Liu B, Nguyen T, Matic LP, Hedin U, Koplev S, Ma L, Franzén O, Ruusalepp A, Schadt EE, Björkegren JLM, **Montgomery SB**, Snyder MP, Quertermous T, Leeper NJ, Miller CL. Functional regulatory mechanism of smooth muscle cell-restricted LMOD1 coronary artery disease locus. PLoS Genet. 2018 Nov 16;14(11):e1007755. doi: 10.1371/journal.pgen.1007755 Role: Aided in HCASMC data production, analysis and manuscript review.

22. Merker JD, Devereaux K, Iafrate AJ, Kamel-Reid S, Kim AS, Moncur JT, **Montgomery SB**, Nagarajan R, Portier BP, Routbort MJ, Smail C, Surrey LF, Vasalos P, Lazar AJ, Lindeman NI. (2018) Proficiency Testing of Standardized Samples Shows Very High Interlaboratory Agreement for Clinical Next-Generation Sequencing-Based Oncology Assays. Arch Pathol Lab Med. 2018 Oct 30. doi: 10.5858/arpa.2018-0336-CP Role: Aided in analyses and manuscript review.

23. Liu, B., Pjanic, M., Wang, T., Gloudemans, M., Rao, A., Castano, V.G., Nurnberg, S.T., Rader, D.J., Elwyn, S., Ingelsson, E. **Montgomery, S.B. [1],** Miller, C.L. [1], Quertermous, T. [1] (2018) Genetic regulatory mechanisms of smooth muscle cells map to coronary artery disease loci. Am J Hum Genet. 2018 Sep 6;103(3):377-388 [1]joint senior authors

24. Rao, A.S., Lindholm, D., Rivas, M.A., Knowles, J.W., **Montgomery, S.B.**, Ingelsson, E. (2018) Large-scale phenome-wide association study of PCSK9 loss-of-function variants demonstrates protection against ischemic stroke, *Circ Genom Precis Med.,* 2018 Jul;11(7). Role: Aided in study design and manuscript review.

25. Wu, Z., Wang, Y., Lim, J., Liu, B., Li, Y., Vartak, R., Stankiewicz, T., **Montgomery, S.B.,** Lu, B. (2018) Ubiquination of ABCE1 by NOT4 in Response to Mitochondrial Damage Links Co-Translational Quanttiy Control to PINK1-Directed Mitophagy, Cell Metabolism, May 18. Role : Aided in RNA-seq analysis and manuscript review

26. Li, Y. [1], Liu, B. [1], Connolly, I.D., Kakusa, B.W., Pan, W., Nagpal, S., **Montgomery, S.B.**, Gephart, M.H. (2018) Recurrently Mutated Genes Differ between Leptomeningeal and Solid Lung Cancer Brain Metastases, Journal of Thoracic Oncology**,** March, 29 [1]joint first authors Role : Aided in analysis and manuscript review.

27. Oláhová, M., Yoon, W.H., 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, Y., Zhao, C., Contrepois, K., Zappala, Z., Frésard, L., Waggott, D., Zink, E.M., Kim, Y.M., Heyman, H.M., Stratton, K.G., Webb-Robertson, B.M.; Undiagnosed Diseases Network, Snyder, M., Merker, J.D., **Montgomery, S.B.**, Fisher, P.G., Feichtinger, R.G., Mayr, J.A., Hall, J., Barbosa, I.A., Simpson, M.A., Deshpande, C., Waters, K.M., Koeller, D.M., Metz, T.O., Morris, A.A., Schelley, S., Cowan, T., Friederich, M.W., McFarland, R., Van Hove, J.L.K., Enns, G.M., Yamamoto, S., Ashley, E.A., Wangler, M.F., Taylor, R.W., Bellen, H.J., Bernstein, J.A., Wheeler, M.T. (2018) Biallelic Mutations in ATP5F1D, which Encodes a Subunit of ATP Synthase, Cause a Metabolic Disorder, AJHG, Mar 1;102(3):494-504. Role: Aided in analysis and manuscript review

28. Gottlieb, A., Daneshjou, R., DeGorter, M., Bourgeois, S., Svensson, P., Wadelius, M., Deloukas, P., **Montgomery, S.B.**, Altman, R. (2017) Cohort-specific imputation of gene expression improves prediction of warfarin dose for African Americans, Genome Medicine, Nov 24;9(1):98 Role : Aided in study design, analysis and manuscript review.

29. Li, X. [1], Kim, Y. [1], Tsang, E.K. [1], Davis, J.R. [1], Damanai, F.N., Chiang, C., Hess, G.T., Zappala, Z., Strober, B.J., Scott, A.J., Li, A., Ganna, A., Bassik, M.C., Merker, J.D., GTEx Consortium, Hall, I.M., Battle, A. *, **Montgomery, S.B.*** (2017) The impact of rare variation on gene expression across tissues, Nature, Oct 11;550(7675):239-243 [1]joint first authors *joint corresponding authors

30. The GTEx Consortium (2017) Genetic effects on gene expression across 44 human tissues. Nature, Oct 11;550(7675):204-213 (**Montgomery,S.B.**, joint corresponding author)

31. The eGTEX Project (2017) Enhancing GTEx by bridging the gaps between genotype, gene expression, and disease. Nature Genetics, Oct 11th, 2017 (**Montgomery,S.B**., joint corresponding author)

32. Ioannidis, N.M., Davis, J.R., DeGorter, M.K., Larson, N.B., McDonnell, S.K., French, A.J., Battle, A., Hastie, T.J., Thibodeau, S.N., **Montgomery, S.B.**, Bustamante, C.D., Sieh, W., Whittemore, A.S. (2017) FIRE : Functional inference of genetic variants that regulate gene expression, Bioinformatics, Aug 24th, 2017 Role : Aided in study design and manuscript review.

33. Merker, J.D., Wenger, A.M., Sneddon, T., Grove, M., Zappala, Z., Frésard, L., Waggot, D., Utiramerur, S., Hou, Y., Smith, K.S., **Montgomery, S.B.**, Wheeler, M., Buchan, J.G., Lambert, C.C., Eng, K.S., Hickey, L., Korlach, J., Ford, J., Ashley, E.A., (2017) Long-read genome sequencing identifies causal structural variation in a Mendelian disease. Genetics in Medicine, Jun 2017 Role : Led transcriptome analysis of Carney case and manuscript review.

34. Knowles, D.A., Davis, J.R., Edgington, H., Raj, A., Favé, M-J., Zhu, X., Potash, J.B., Weissman, M.M., Shi, J., Levinson, D.F., Awadalla, P., Mostafavi, S., **Montgomery, S.B.**[*], Battle, A[*] (2017) Allele-specific expression reveals interactions between genetic variation and environment [*]joint corresponding authors, Nature Methods, AOP May 22, 2017,

35. Steri, M., Orrù, V., Idda, M.L., Pitzalis, M., Pala, M., Zara, I., Faà, V., Floris, M., Deiana, M., Asunis, I., Sidore, C., Porcu, E., Mulas, A., Marongiu, M., Serra, V., Lobina, M., Piras, M.G., Marongiu, M., Sole, G., Busonero, F., Maschio, A., Cusano, R., Cuccuru, G., Deidda, F., Poddie, F., Farina, G., Dei, M., Lai, S., Virdis, F. Olla, S., Satta, M.A., Pani, M, Cocco, E., Frau, J., Coghe, G., Lorefice, L., Fenu, G., Ferrigno, P., Ban, M., Barizzone, N., Leone, M., Guerini, F.R., Piga, M., Firinu, D., Kockum, I., Bomfim, I.L., Olsson, T., Alfredsson, L., Suarez, A., Carreira, P., Castillo-Palma, M.J., Marcus, J.H., Congia, M., Angius, A., Melis, M., Gonzalez, A., Riquelme, M.E.A., Da Silva, B.M., Marchini, M., Danieli, M.G., Del Giacco, S., Mathieu, A., Pani, A., **Montgomery, S.B.**, Rosati, G., Hillert, J., Sawcer, S., D´Alfonso, S., Todd, J.A., Novembre, J., Abecasis, G.R., Whalen, M.B., Marrosu, M.G., Meloni, A., Sanna, S., Gorospe, M., Schlessinger, D., Fiorillo, E., Zoledziewska, M., Cucca, F. (2017) Evolutionarily selected overexpression of the cytokine BAFF raises autoimmunity risk, [1]joint first authors NEJM, Apr 27;376(17):1615-1626. Role : Contributed to eQTL analyses of BAFF variants.

36. Pala, M.,[1] Zappala, Z.[1], Marongiu, M., Li, X., Davis, J.R., Cusano, R., Crobu, F., Kukurba, K.R., Reiner, F., Berutti, R., Piras, M.G., Mulas, A., Zoledziewska, M., Marongiu, M., Busonero, F., Maschio, A., Steri, M., Sidore, C., Sanna, S., Fiorillo, E., Battle, A, Novembre, J., Jones, C., Angius, A., Abecasis, G.R., Schlessinger, D., Cucca, F.[*], **Montgomery, S.B.**[*] (2017) Population and individual effects of non-coding variants inform genetic risk factors [1]joint first authors [*]joint corresponding authors, Nature Genetics, May;49(5):700-707

37. Chiang, C., Scott, A.J., Davis, J.R., Tsang, E.K., Li, X., Kim, Y., Hadzic, T., Damani, F., Ganel, L., GTEx Consortium, **Montgomery, S.B.**, Battle, A., Conrad, D.F., Hall, I.M. (2017) The impact of structural variation on human gene expression, Nature Genetics, May;49(5):692-699. Role : Contributed to study design, analysis of rare structural variants and manuscript review.

38. 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., Léger, D., Frost, L.D., **Montgomery, S.B.**, Lewis, S.M., Eskiw, C., Dellaire, G. (2017) PML nuclear bodies contribute to the basal expression of the mTOR inhibitor DDIT4. Sci Rep. 2017 Mar 23;7:45038. Role : Conducted transcriptome sequencing and analysis for PML knockdowns.

39. Kernohan, K.D.[1], Frésard, L.[1], Zappala, Z.[1], Hartley, T., Smith, K.S., Wagner, J., Xu, H., McBride, A., Bourque, P.R., Consortium CR, Bennett, S.A., Dyment, D.A., Boycott, K.M.[*], **Montgomery, S.B.**[*], Warman Chardon, J.[*] (2017) Whole-transcriptome sequencing in blood provides a diagnosis of spinal muscular atrophy with progressive myoclonic epilepsy**,** [1]joint first authors [*]joint senior authors Hum Mutat. 2017 Jun;38(6):611-614.

40. 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) Small RNA sequencing in cells and exosomes identifies eQTLs and 14q32 as a region of active export, G3, Jan 5;7(1):31-39.

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92. Griffith, O.L.[1], **Montgomery, S.B.[1],** Bernier, B., Chu, B., Aerts, S., Sleumer, M.C., Bilenky, M., Haeussler, M., Griffith, M., Gallo, S.M., Giardine, B., Mahony, S., Hooghe, S., Van Loo, P., Blanco, E., Ticoll, A., Lithwick, S., Portales-Casamar, E., Donaldson, I.J., Robertson, A.G., Wadelius, C., De Bleser, P., Vlieghe, D., Halfon, M.S., Wasserman, W.W., Hardison, R., Bergman, C.M., Jones, S.J.M. The Open Regulatory Annotation Consortium, ORegAnno: an open-access community-driven resource for regulatory annotation. Nucleic Acids Research. 2008, Jan ;36 :D107-13 [1]joint first authors.

93. 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.B.,** Tavare, S., Deloukas, P. and Dermitzakis, E.T., Population genomics of human gene expression. Nature Genetics, 2007, 39(10):1217-24 Role : Contributed analysis.

94. **Montgomery, S.B.,** Griffith, O.L., Scheutz, J.M., Brooks-Wilson, A., Jones, S.J., A Survey of Genomic Properties for the Detection of Regulatory Polymorphisms. PLoS Computational Biology, 2007, 8;3(6):e106

95. **Montgomery, S.B.[1],** Griffith, O.L.[1], Sleumer, M.C., Bergman, C.M., Bilenky, M., Pleasance, E.D., Prychyna, Y., Zhang, X., Jones, S.J., ORegAnno: an open access database and curation system for literature-derived promoters, transcription factor binding sites and regulatory variation. Bioinformatics, 2006, 1;22(5):637-50 [1]joint first authors

96. Robertson, A.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, M.C., Pan, W., Pleasance, E.D., Chuang, M., Hao H., Li, Y.Y., Robertson, N., Fjell, C., Li, B., **Montgomery, S.B.,** Astakhova, T., Zhou, J., Sander, J., Siddiqui, A.S. and Jones, S.J., cisRED: A database system for genome scale computational discovery of regulatory elements. Nucleic Acids Research, 2006, 1;34 (Database issue):D68-73. Role : Contributed to study design and analysis.

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

98. **Montgomery, S.B.,** Astakhova, T., Bilenky, M., Birney, E., Fu, T., Hassel, M., Melsopp, C., Rak, M., Robertson, A.G., Sleumer, M.C., Siddiqui, A.S., and Jones, S.J., Sockeye: A 3D Environment for Comparative Genomics. Genome Research, 2004, 14:956-962 (Sockeye also featured on the cover of Linux Journal)

99. 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, D., Girn, N., Griffith, O.L., Leach, S.R., Mayo, M., McDonald, H., **Montgomery, S.B.,** Pandoh, P.K., Petrescu, A.S., Robertson, A.G., Schein, J.E., Siddiqui, A.S.,

Smailus, D.E., Stott, J.M., Yang, G.S., Plummer, F., Andonov, A., Artsob, H., Bastien, N., Bernard, K., Booth, T.F., Bowness, D., Czub, M., Drebot, M., Fernando, L., Flick, R., Garbutt, M., Gray, M., Grolla, A., Jones, S., Feldmann, H., Meyers, A., Kabani, A., Li, Y., Normand, S., Stroher, U., Tipples, G.A., Tyler, S., Vogrig, R., Ward, D., Watson, B., Brunham, R.C., Krajden, M., Petric, M., Skowronski, D.M., Upton, C., Roper, R.L., The Genome sequence of the SARS associated coronavirus. Science, 2003, 300(5624):1399-404 Role : contributed to analysis

PEER-REVIEWED REVIEWS*

***Underlined highlights myself or people in my laboratory.**

1. Liu, B, **Montgomery, SB.** (2019) Identifying causal variants and genes using functional genomics in specialized cell types and contexts. Human Genetics. Jul 17. doi: 10.1007/s00439-019-02044-2
2. Frésard L, **Montgomery SB.** (2018) Diagnosing rare diseases after the exome. Cold Spring Harb Mol Case Stud. 2018 Dec 17;4(6). pii: a003392. doi: 10.1101/mcs.a003392.
3. Ritchie, M.D.*, Davis, J.R., Aschard, H., Battle, A., Conti, D., Mengmeng, D., Eskin, E., Fallin, M.D., Hsu, L., Kraft, P., Moore, J.H., Pierce, B.L., Bien, S., Thomas, D.C., Wei, P., **Montgomery, S.B.*** (2017) Incorporation of Biological Knowledge into the Study of Gene-Environment Interactions, *joint corresponding authors, AJE, Volume 186, Issue 7, 1 October 2017, Pages 771–777
4. McAllister, K., Mechanic, L.E., Amos, C., Aschard, H., Blair, I., Chatterjee, N., Conti, D., Gauderman, W.J., Hsu, L., Hutter, C.M., Jankowska, M., Kerr, J., Kraft, P., **Montgomery, S.B.**, Mukherjee, B., Papanicolaou, G.J., Patel, C.J., Ritchie, M.D., Ritz, B.R., Thomas, D.C., Wei, P., Witte, J.S. (2017) Current Challenges and New Opportunities for Gene-Environment Interaction Studies of Complex Diseases, AJE, Volume 186, Issue 7, 1 October 2017, Pages 753–761 Role: Contributed to NIEHS overview manuscript of GxE analyses.
5. Zappala, Z., **Montgomery, S.B.** (2016) Non-Coding Loss-of-Function Variation in Human Genomes, Human Heredity 2016;81(2):78-87
6. Kukurba, K.R., **Montgomery, S.B.** (2015) RNA-Sequencing and Analysis, Cold Spring Harb Protoc., Apr 13;2015(11):951-69
7. Battle, A., **Montgomery, S.B.**, (2014) Determining causality and consequence of expression quantitative trait loci, Human Genetics, 2014, Jun ;133(6) :727-35
8. Miller, C.L., Assimes, T.L., **Montgomery, S.B.,** Quertermous, T., Investigation of causal genetic mechanisms for coronary heart disease genome-wide associations, Curr Atheroscler Rep, 2014;16:406
9. Li, X., **Montgomery, S.B.**, Detection and impact of rare regulatory variants in human disease, Frontiers in Statistical Genetics and Methodology, May 31 ;4 :67
10. **Montgomery, S.B.,** Dermitzakis, E.T., From Expression QTLs to Personalized Transcriptomics. Nature Reviews Genetics, 2011, Apr ; 12(4) :277-82
11. **Montgomery, S.B.,** Dermitzakis, E.T.,The resolution of the genetics of gene expression. Human Molecular Genetics, 2009, R(2):R211-5

NON-PEER-REVIEWED PUBLICATIONS*

***Underlined highlights myself or people in my laboratory.**

1. Karczewski, K.J., **Montgomery, S.B.**, The performance of genomic medicine Genome Biology, 2013, 14:316
2. Morin, R. **Montgomery, S.B.**, Cancer Transcriptome Sequencing and Analysis (Chapter 5), Cancer Genomics: From Bench to Personalized Medicine, 2013
3. Ning, Z., **Montgomery, S.B.,** Out of the sequencer and into the wiki as we face new challenges in genome informatics, Genome Biology, 2010, 11:308
4. **Montgomery, S.B.,** Kasaian, K., Jones, S.J.M., Griffith, O.L. Annotating the Regulatory Genome (Chapter 20). Computational Biology of Transcription Factor Binding. Methods Mol Biol. 2010;674:313-49.
5. **Montgomery S.B.** Current computational methods for prioritizing candidate regulatory polymorphisms. Methods in Molecular Medicine. Humana Press. 2009, 569 :89-114

PHD AND POSTDOCTORAL TRAINEES

Konrad Karczewski, Ph.D. student, BMI, co-mentor Michael Snyder, now faculty at MGH	2012-2013
Olga Sazonova, Ph.D., Postdoctoral Researcher, now a scientist at 23andme	2013-2014
Hoon Cho, Undergraduate Student, now a grad student at MIT	2012-2013
Mauro Pala, Ph.D., Visiting Graduate Student, now a scientist CNRS (Sardinia)	2012-2013
Tracy Nance, Ph.D., Postdoctoral Researcher, now at Guardant Health	2012-2014
Kim Kukurba, Ph.D. student, Genetics, now at Raytheon	2012-2015

Zach Zappala, Ph.D. student, Genetics, now at Vertex Pharmaceuticals	2013-2017
Joe Davis III, Ph.D. student Genetics, now at Cardinal Analytics	2013-2017
Emily Tsang, Ph.D. student, BMI, now at Guardant Health	2013-2018
Brunilda Balliu, Ph.D., Postdoctoral Researcher, now at UCSD as a Fellow	2015-2018
Bosh Liu, Ph.D. student, Biology, now at Baidu	2014-2019
Xin Li, Ph.D., Postdoctoral Researcher, faculty in Shanghai	2012-2019
Laure Fresard, Ph.D., Postdoctoral Researcher, CEHG Fellow, now at Invitae	2015-2020
Craig Smail, Ph.D. student, BMI, faculty at Children's Mercy (10/20)	2017-2020
Abhiram Rao, Ph.D. student, Bioengineering, co-mentor Erik Ingelsson, now at Tempus (9/20)	2016-2020
Marianne DeGorter, Ph.D., Postdoctoral Researcher, Banting Fellow	2013-
Daniel Nachun, Ph.D., Postdoctoral Researcher, co-mentor Sidd Jaiswal	2018-
Nathan Abell, current Ph.D. student, Genetics	2016-
Michael Gloudemans, current Ph.D. student, BMI	2016-
Nikki Teran, current Ph.D. student, Genetics	2016-
Nicole Ferraro, current Ph.D. student, BMI	2017-
Matt Durant, current Ph.D. student, Genetics, co-mentor Ami Bhatt	2017-
Olivia de Goede, current Ph.D. student, Genetics, co-mentor Karla Kirkegaard	2017-
Nicole Gay, current Ph.D. student, Genetics	2018-
Kameron Rodrigues, current Ph.D. student, Immunology, co-mentor Sidd Jaiswal	2018-
Tiffany Eulalio, current Ph.D. student, BMI	2019-
Pagé Goddard, current Ph.D. student, Genetics	2019-
Emily Greenwald, current Ph.D. student, Genetics, co-mentor Andrew Fire	2019-
Rachel Ungar, current Ph.D. student, Genetics	2019-

Ph.D. Rotation students : Rhea Richardson (Genetics), Michael Haney (Genetics), Michael Sikora (Genetics), Erin Mitsunaga (Genetics), David Moskowitz (BMI), Jessica Ribado (Genetics), Eli Moss (Genetics), Rachel Agoglia (Genetics), Siming Zhang (Genetics), Cameron Prybol (Genetics), Nikki Teran (Genetics), Tyler Shimko (Genetics), Anna Shcherbina (BMI), Mohana Moorthy (CS Masters student), Candace Liu (Immunology), Daniel Cotter (Genetics), Matthew Aguirre (BMI), Courtney Smith (Genetics), Michael Hayes (Genetics), Jarod Rutledge (Genetics)

Computer Science advisees and RAs: Mira Noura Moufarrej (MS), Preston Ng (MS), Cooper Douglas Raterink (MS), Luis Govea (MS), Anirudh Joshi (MS), Gauri Prasad (MS), Hoon Cho (Undergrad RA), Mohana Moorthy (MS/RA)

Electrical Engineering advisees and RAs : Vikranth Dwaracherla (PhD/RA)

BMI advisees and RAs : Sheun Aluko (MS), Emily Guthrie (Co-term), Shuyu (Chloe) He (MS), Derek Jow (MS), Sandra Kong (Co-term), Kaitlyn Lagattuta (Co-term), Yousuf Khan (MS), Samson Mataraso (PhD), Dylan Peterson (MS), Katherine Shi (HCP-MS), Yosuke Tanigawa (PhD), Juan Manuel Zambrano Chaves (PhD), Mahdi Moqri (MS/RA)

Stanford Outreach Programs : Tahla Lone (Nicole Gay/undergrad/SSRP), Jeremy Tien, SIMR/high-school student, Jessica L. Saal Award, Eric Wu, SIMR/high-school student, Tony Jiang, SIMR/high-school student.

Thesis advisory/qualifying/defense commitees: Michael Sikora (Genetics), Julian Homburger (Genetics), Nora Yucel (Genetics), Helio Costa (Genetics), Alicia Martin (Genetics), Gokul Ramaswami (Genetics), Sofia Kyriazopoulou-Panagiotopoulou (CS), Oana Ursu (BMI), Shaila Musharoff (Genetics), Trevor Martin (Biology), Yi Liu (BMI), David Poznik (BMI), Azad Raiesdana (Medicine), Kimberley McManus (Genetics), Zoe Assaf (Biology), Ryosuke Kita (Biology), Linda Szabo (BMI), Tyler Shimko (Genetics), Rachel Agoglia (Genetics), Emily Glassberg (Biology), Ariel Yablonovitch (Genetics), Jessica Ribado (Genetics), Weizhuang Zhou (Bioengineering), Karthik Jagadeesh (Computer Science), Stephanie Nevins (Genetics), Ben Siranosian (Genetics), Roshni Patel (Genetics), Daniel Cotter (Genetics), Alex Kern (Genetics), Siming Zhang (Genetics)

AWARDS AND SPECIAL RECOGNITION

- 2019 Highly Cited Researchers by Web of Science/Clarivate Analytics
- 2019 ASHG Early Career Award
- 2011 Genome Technology Magazine Rising PI (December 2011 issue)

- 2010 A-Star New Investigator Award (not accepted, conditioned on move)
- NSERC (Natural Sciences and Engineering Research Council) postdoctoral fellowship.
- NSERC UK Millennium Research Award.
- EMBO (European Molecular Biology Organization) Long-term postdoctoral fellowship.
- Co-PI on Genome BC funding for “The Regcreative Workshop” in Ghent, Belgium.
- Co-PI on Genome Canada funding for “The Regcreative Workshop” in Ghent, Belgium.
- BC Cancer Agency Lloyd Skarsgard Research Excellence Prize (awarded annually to single BC Cancer Agency graduate student)
- NSERC PGS-Doctoral Scholarship.
- Invited to attend “Encoding Information in DNA Sequences” conference in Okinawa, Japan (40 students of 400 selected)
- BCNET Best Overall winner for Chinook project.
- MSFHR (Michael Smith Foundation for Health Research) Senior Studentship Award.
- UBC PhD Tuition Fee Award.
- EBI Travel Grant for EnsEMBL Developers Day.

PROFESSIONAL AND VOLUNTEER SERVICE

Editorial service: American Journal of Human Genetics, Associate Editor, 2018-2021
PLoS Computational Biology, *ad hoc* Guest Editor, 2014-
PLoS Genetics, *ad hoc* Guest Editor, 2012-
BMC Genomics, Associate Editor, 2012-2015
eLife, *ad hoc* Guest Editor, 2016-

NIH Study Sections: Meeting 2021/01 ZAI1 TCB-I-J2 (NIAID)
Meeting 2020/10 ZDE1-NB-03 (NIDCR)
Meeting 2020/10 ZDK1-GRB-7-O4 (NIDDK)
Meeting 2020/05 ZDK1-GRB-N-M3 (NIDDK)
Meeting 2020/05 ZDE1-NB-21 (NIDCR)
Meeting 2020/05 ZAI1-MMF-I-M2 (NIAID)
Meeting 2020/01 ZAI1-LAR-I-J2 (NIAID)
Meeting 2019/10 ZDK1-GRB-7-O4 (NIDDK)
Meeting 2019/10 DSR (NIDCR)
Meeting 2019/05 ZDK1-GRB-J-M2 (NIDDK)
Meeting 2019/05 ZDK1-GRB-7-M4 (NIDDK)
Meeting 2019/05 ZDE1-NB-14 (NIDCR)
Meeting 2019/01 ZAI1-PTM-I-J1 (NIAID)
Meeting 2018/10 ZDK1 GRB-7 (O4) 1 (NIDDK)
Meeting 2018/10 ZAI1 JA-I (S1) 2 (NIAID)
Meeting 2018/10 DDK-C 1 (NIDDK)
Meeting 2018/05 ZGM1-TRN-9-MR (NIGMS)
Meeting 2018/05 ZDK1-GRB-S-M6 (NIDDK)
Meeting 2018/01 GHD (CSR)
Meeting 2017/10 GVE (CSR)
Meeting 2017/05 ZDK1-GRB-N-M2 (NIDDK)
Meeting 2017/05 ZRG1-ETTN-G-02 (CSR)
Meeting 2017/01 GVE (CSR)
Meeting 2016/05 ZAI1-LAR-I-M1 (NIAID)
Meeting 2016/01 ZHG1-HGR-M-J1 (NHGRI)
Meeting 2015/05 ZGM1-PPBC-Y-PG (NIGMS)
Meeting 2014/01 ZAI1-LAR-I-S1 (NIAID)
Meeting 2013/05 ZAI1 LAR-I-M2 (NIAID)

Grant reviewer: NSF GRFP, Genetics, Genomics and Proteomics Panel, 2020
Genome Canada GAPP Pitch Review, 2019
Genome Alberta RP3 Review, 2019
Genome Canada GAPP Pitch Review, 2018
Wellcome Trust (UK), Senior Investigator Awards, 2018
Genome Canada, Disruptive Innovation in Genomics Competition, 2018
Wellcome Trust/DBT India Alliance, Fellowships, 2018
Marsden Fund, Royal Society of New Zealand, 2017
NASA, TRI Omics Study Section, 2017
Genome Canada, Disruptive Innovation in Genomics Competition, 2015

Research Foundation Flanders (Belgium), 2015
Medical Research Council (UK), 2014
Israeli Science Foundation, 2014
Stanford Child Health Research Institute, 2012-
Genome Quebec / Genome Canada, 2012
Natural Sciences and Engineering Research Council, 2013
Ministry of Health, Singapore, 2011

Ad hoc reviewer: AJHG, Bioinformatics, Cell Systems, Cell Stem Cell, Database, eLife, Genome Biology and Evolution, Frontiers in Genetics, Genome Research, Human Genetics, Human Molecular Genetics, Human Mutation, Molecular Systems Biology, Nature, Nature Biotechnology, Nature Genetics, Nature Immunology, Nature Reviews Genetics, Nucleic Acids Research, PLoS Comp Bio, PLoS Genetics, PLoS One, PNAS, Science

Other service:
ASHG Program Committee (2021 – 2023)
Elected to Stanford Faculty Senate (2020 – 2022)
Genome Canada Future of Genomics Technology SAB member, 2020
AGBT Organizing Committee 2020
Stanford BMI Executive Committee (2019 -)
BASE Faculty Search Committee
MCHRI Grant Reviewer
BIRS 2017 organizer http://www.birs.ca/events/2017/5-day-workshops/17w5131
BIRS 2015 organizer https://www.birs.ca/events/2015/5-day-workshops/15w5142
BIRS 2013 organizer https://www.birs.ca/events/2013/5-day-workshops/13w5083
European Conference of Computational Biology 2012, 2014, 2016 – PC member
ASHG Mentor/Trainee Luncheon 2015, 2016, 2017, 2018, 2019
Stanford Genetics Graduate Student Admission committee
Stanford Center for Genomics and Personalized Medicine Advisory Committee
Stanford Medicine and Hospitals Genome Sequencing Advisory Committee
Department Senator, Stanford School of Medicine
Faculty of 1000, Associate Member, 2011-2012
Education outreach, STIMULUS Cambridge
Conference organizer, The Regcreative Jamboree, Ghent Belgium
Education outreach, UBC Let's Talk Science Partnership Program
Conference organizer, Vancouver Bioinformatics User Group, www.vanbug.org.
Education outreach, Java and bioinformatics article series for O'Reilly and Associates. "Java for Bioinformatics" and "Java APIs for Bioinformatics"

Group memberships: ClinGen, NESCENT cisevol working group, 1000 Genomes Project, MoTrPAC, ENCODE4, TOPMED, Hapmap 3 Consortium, GEUVADIS project, GTEx project, MuTHeR project, SardiNIA project, Alspac analysis group and Canadian Data Integration Centre.

SAB/Consulting: Prime Genomics (until 2019), MyOme Ltd. (2020-), GLG(2019-)

TEACHING ACTIVITIES

- Instructor for BIOS201 course since 2013. ~400-500 students have been instructed on DNA sequence analysis and quality control, variant calling and differential analyses through hands-on activities. Current workshops are here: https://github.com/zaczap/bios201
- Instructor for BIOS 200 Foundations course in 2018, 2019, 2020 (upcoming). Students are guided in developing research proposals and talks.
- Sardinia Summer School on Human Genetics: Instructor 6 times on functional genomics data analysis.
- Co-organize with Julia Salzmann BMI's lecture series from affiliated faculty.
- Stanford Biosciences Grant Writing Workshops aiding trainees in writing F32/K applications.
- Guest lecturer for GENE203, BIOMED205, MI215, NEPR202, Genetics/DevBio Training Camp and GENE210

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Montgomery, S.B. (2002) BindingSite Application. Computer Application and Code used for Comparing Multiple Strands of DNA. Canadian Intellectual Property Office Copyright. File No. 418724

Montgomery, S.B. and Smail, C. 2020. US Provisional Patent Ref: 62/934,892. Estimation of phenotypes using genetic data.

PRESENTATIONS/INVITED MEETINGS (since 2011)

University of Washington, Seattle, WA (Invited Speaker) 2019
BC Genome Science Center, Vancouver, BC (Invited Speaker) 2019
Washington University, St Louis, MO (Invited Speaker) 2019
Sardinia Summer School in Genomics, Pula, Italy (Invited Speaker) 2019
AGBT Precision Medicine, La Jolla, CA (Invited Speaker) 2019
AGBT, Marco Island, FL (Invited Speaker) 2019
Frontiers in Cardiovascular Science Seminar, Stanford, CA (Invited Speaker) 2019
NHLBI TOPMed F2F Meeting, Washington, DC (Invited Speaker) 2018
Sidra Medicine, Doha, Qatar (Invited Speaker) 2018
Precision Genomics Midwest, Cincinnati, USA (Invited Speaker) 2018
Winter Human Genetics Seminar Series, UC Davis, USA (Invited Speaker) 2018
Mount Sinai, Dept of Genetics and Genomic Sciences (Invited Speaker) 2017
University of Pittsburgh, Dept of Human Genetics (Invited Speaker) 2017
ADA 77th Scientific Meeting, San Diego, USA (Invited Speaker) 2017
BioConductor Meeting, Stanford, USA, (Invited Speaker) 2016
Sardinia Summer School in Genomics , Pula, Italy (Invited Speaker) 2016
FIMM Think Different Seminar Series, Helsinki, Finland (Invited Speaker) 2016
2nd Barbados Workshop on Population Epigenomics (Invited Speaker) 2016
EMBL|Stanford Conference : Personalized Health, Heidelberg, Germany (Invited Speaker) 2015
American Society of Human Genetics Meeting, Baltimore, USA (Invited Speaker) 2015
BIRS, Banff, Canada (Conference Organizer, Speaker) 2015
Winter School in Mathematics and Computational Biology, Australia (Invited Speaker) 2015
Sardinia Summer School in Genomics, Pula, Italy (Invited Speaker) 2015
Symposium on Advances in Genomics, Epi and Stats, UPenn, USA (Invited Speaker) 2015
Cartagene Conference, Montreal, Canada (Invited Speaker) 2015
"Rare regulatory variation in individuals, families and populations"
UCSF Institute of Human Genetics Seminar, San Francisco, USA (Invited Speaker) 2015
University of Chicago Human Genetics Seminar, Chicago, USA (Invited Speaker) 2015
CSHL Systems Biology of Gene Regulation, Rio Mar, Puerto Rico (Speaker) 2015
European Union FP7 Syscol Workshop, Wolfenbuttel, Germany (Invited Speaker) 2014
Wellcome Trust Genome Informatics Meeting, Cambridge, UK (Speaker) 2014
ICSB Regulatory Genetics SIG, Boston, USA (Invited Speaker) 2014
Big Data in Biomedicine, San Francisco, USA (Session host) 2014
TriCon Molecular Medicine, San Francisco, USA (Invited Speaker) 2014
UC Davis Guest Lecturer, Davis, USA (Invited Speaker) 2013
UC Berkeley Guest Lecturer, Berkeley, USA (Invited Speaker) 2013
CSHL Precision Medicine, Cold Spring Harbor, USA (Speaker) 2013
"Interpreting the impact of rare, deleterious and loss-of-function variants from transcriptome data"
American Society of Human Genetics Meeting, San Francisco, USA (Invited Speaker) 2013
International Genetic Epidemiology Society, Chicago, USA (Invited Speaker) 2013
Sardinia Summer School in Genomics, Pula, Italy (Invited Speaker) 2013
Canadian Institute for Advanced Research, Banff, Canada (Invited Speaker) 2013
BIRS, Banff, Canada (Conference Organizer, Speaker) 2013
Sardinia Summer School in Genomics , Pula, Italy (Invited Speaker) 2012
International Conference on Systems Biology, Toronto, Canada (Invited Speaker) 2012
"Analysis of gene expression patterns in a single extended family"
Capita Select in Complex Disease Analysis, Liege, Belgium (Invited Speaker) 2012
Personalized Medicine World Congress, Mountain View, USA (Invited Speaker) 2012
Swedish Medical Genetics Society, Uppsala, Sweden (Invited Speaker) 2011
"Transcriptome sequencing and diagnostics"
International Conference on Systems Biology, Mannheim, Germany (Invited Speaker) 2011
Sardinia Summer School in Genomics, Pula, Italy (Invited Speaker) 2011
European Society of Human Genetics, Amsterdam, Netherlands (Invited Speaker) 2011
Gordon Conf. on Quantitative Genetics and Genomics, Galveston, USA (Invited Speaker) 2011

ADVANCED TRAINING

Stanford Grant Writing Course (R01 Countdown Pilot)
CITI Group 1 training certificate #8919278

NIH Protecting Human Research Participants certificate # 422835
UBC Advanced Molecular Biology Laboratories “Molecular Biology Techniques Workshop”
UBC Professional Training Workshop “Leadership and Management Workshop”

MEDIA*

*includes media where I was asked to provide a comment

Healthline, How doctors from across the globe saved an infant with just months to live. 2020
https://www.healthline.com/health-news/how-doctors-saved-an-infant-with-3-weeks-to-live
GenomeWeb, RNA-Seq in Blood Cell Line Enables Molecular Diagnosis. 2020
https://www.genomeweb.com/molecular-diagnostics/rna-seq-blood-cell-line-enables-molecular-diagnosis-neurodevelopmental
ASHG Press, https://www.ashg.org/press/201907-EarlyCareer-Award.shtml 2019
Globe and Mail, Genetic study of Quebec residents finds air pollution trumps ancestry 2018
https://www.theglobeandmail.com/news/national/genetic-study-of-quebec-residents-finds-air-pollution-trumps-ancestry/article38217989/?click=sf_globe
Gizmodo, Unprecedented Video Shows How DNA Is Organized in Real Time 2018
https://gizmodo.com/unprecedented-video-shows-how-dna-is-organized-in-real-1823211811
The Scientist, Massive Transcription Catalog Outlines the Influence of Human Genetic… 2017
http://www.the-scientist.com/?articles.view/articleNo/50602/title/Massive-Transcription-Catalog-Outlines-the-Influence-of-Human-Genetic-Variation/
Stanford Medicine News, Tissue-specific gene expression uncovered, linked to disease 2017
http://med.stanford.edu/news/all-news/2017/10/tissue-specific-gene-expression-uncovered-linked-to-disease.html
AlzForum, Gene Expression Map of Human Body Gives Value to Variants 2017
http://www.alzforum.org/news/research-news/gene-expression-map-human-body-gives-value-variants
GenomeWeb, Studies Elucidate Impact of Genetic Variants on Gene Expression in Human… 2017
https://www.genomeweb.com/sequencing/studies-elucidate-impact-genetic-variants-gene-expression-human-tissues
Cosmos, Cracking the mystery of human variation, at last 2017
https://cosmosmagazine.com/biology/cracking-the-mystery-of-human-variation-at-last
MIT Technology Review, A DNA App Store Is Here But Proceed With Caution 2017
https://www.technologyreview.com/s/608313/a-dna-app-store-is-here-but-proceed-with-caution/
GenomeWeb, Parody Genomics 2017
https://www.genomeweb.com/scan/parody-genomics
Gizmodo, Scientists Push Back Against Booming Genetic Pseudoscience Market 2017
http://gizmodo.com/scientists-push-back-against-booming-genetic-pseudoscie-1796923059
The Scientist, Abundant Sequence Errors in Public Databases 2017
http://www.the-scientist.com/?articles.view/articleNo/48510/title/Abundant-Sequence-Errors-in-Public-Databases/
Stanford News, 10.5 million awarded to researchers to work on DNA encyclopedia 2017
https://med.stanford.edu/news/all-news/2017/02/10-million-awarded-to-researchers-to-work-on-dna-encyclopedia.html
Stanford News, NIH awards 26.4 million to Stanford researchers for physical activity study 2016
http://med.stanford.edu/news/all-news/2016/12/researchers-awarded-more-than-26-million-for-activity-study.html
RadioNZ, This Way Up, How Genes Define Us 2016
http://www.radionz.co.nz/national/programmes/thiswayup/audio/201795485/how-genes-define-us
BBC News, Are fitbands the future of genetic research? 2016
http://www.bbc.com/news/science-environment-35411685
GenomeWeb, Stanford Team to Use Targeted RNA-seq for Allele-Specific Expression 2014
https://www.genomeweb.com/sequencing/stanford-team-use-targeted-rna-seq-allele-specific-expression-analysis-gtex-samp
Genome Technology Magazine, Sixth Annual Young Investigators 2011
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GRANTS

ACTIVE

R01DK120565 (Knowles, Joshua, PI) 09/13/2019-05/31/2024 0.30 calendar
National Institutes of Health
Characterization of Novel Insulin Resistance Genes by Gene Editing, High-throughput Phenotyping and in vivo Studies
Major Goal: Establish causal genes and mechanisms of action for novel genes involved in development of insulin resistance, by combining a range of innovative methods including high-throughput gene perturbations followed by single-cell transcriptomics, in vitro and in vivo experiments, to characterize loci established using human genetics.
Role : Co-investigator. Aiding with various statistical and computational analyses and methods development.

R01AG066490 (Montgomery, Stephen B., PI) 02/01/2020-01/31/2025 1.80 calendar
National Institutes of Health
Mapping Molecular and Phenotypic Interactions in Alzheimer's Disease
Major Goal: Develop novel computational methods to combine large data sets of genetic and phenotypic data, as well as functional genomics, to further elucidate biological processes, pathways, and cell types leading to Alzheimer's disease.

1R01HL142015 (Montgomery, Stephen B., PI) 05/01/2018-04/30/2021 NCX 1.20 calendar
National Institutes of Health
Integrative multi-omics in whole genome studies of HLBS disorders
Major Goals: Computational analysis of rare variants and gene-by-environment variants in NHLBI's TOPMED project.

1U01HG009431 (Pritchard, Jonathan) 02/01/2017-01/31/2021 0.60 calendar
National Institutes of Health
Decoding the Regulatory Architecture of the Human Genome Across Cell Types, Individuals and Disease
Major Goals: This project focuses on analysis of ENCODE data to identify regulatory variants involved in human disease.
Role: Co-investigator. I helped write this grant. My laboratory is focused on integrating ENCODE data to aid in prediction of impactful rare regulatory variants

1U24DK112348 (MPI: Snyder, M./Montgomery, S.) 12/13/2016-11/30/2022 3.0 calendar
National Institutes of Health
Stanford/Salk MoTrPAC Site for Genomes, Epigenomes and Transcriptomes
Major Goals: The Stanford/Salk MoTrPAC site is designed to provide genomes, transcriptomes and epigenomes for the MoTrPAC to elucidate molecular changes in response to physical activity.

1R01HL145708 (Quertermous, Thomas) 1/18/2019-12/31/2022 0.60 calendar
National Institutes of Health
LncRNA Transcriptional Mechanisms of Coronary Artery Disease Risk
Major Goals: Montgomery lab: Identify and validate lncRNA eQTLs influencing CAD using mmPCR-seq approach.
Role: Co-investigator. I helped write this grant. My lab is focusing on lncRNA QTLs in GWAS.

1U01HG010218 (Ashley, Euan) 09/21/2018-08/31/2022 0.24 calendar
National Institutes of Health
Center for Undiagnosed Diseases at Stanford (Undiagnosed Disease Network Phase II)
Major Goals: Transition UDN Centers toward sustainability by: 1) facilitating undiagnosed disease diagnoses, 2) improving diagnostic rates through novel approaches to data analysis/integration, 3) exploring underlying disease mechanisms to accelerate therapeutic drug discovery. *Role: Co-investigator.*

0258-A061-4609 (Montgomery, Stephen B., Co-I) 04/01/2019-03/31/2021 2.16 calendar
Defense Advanced Research Project Agency / Icahn School of Medicine at Mount Sinai
WMD ECHO Detector

Major Goal: Build ESP and associated bioinformatics algorithms to identify militarily relevant exposures and develop epigenetics analysis and exposure identification platform.

COMPLETED

1R01DK107437 (Quertermous, Thomas) 01/01/2016-12/31/2019 0.84 calendar
National Institutes of Health
Molecular Mechanisms of Insulin Resistance Associated Loci
Major Goals: The main goal of this project is to apply genome, transcriptome and epigenome sequencing to identify causal regulatory variants underlying insulin resistance-associated variation.
Role: Co-investigator. My laboratory is aiding in analysis of genome and transcriptome data to identify causal regulatory variants underlying insuline resistance-associated variation.

1U01HG00908001 (Bustamante, Carlos D.) 05/02/2016-03/31/2020 0.60 calendar
National Institutes of Health
Center for Multi- and Trans-ethnic Mapping of Mendelian and Complex Diseases
Major Goals: The goal of this GSPAC Center grant is to analyze common and rare variants in 250,000 genomes across a range of ethnicities. Dr. Montgomery shares a postdoc with Dr. Bustamante on this grant.
Role: Co-investigator. I helped write a small part of this grant. My laboratory is focused on multi-ethnic eQTL analyses in African populations and their utility for fine-mapping causal alleles.

1R01HG008150 (Montgomery, Stephen B., PI) 08/01/2015-06/30/2019 1.20 calendar
National Institutes of Health
Predicting Casual Non-Coding Variants in a Founder Population
Major Goals: The main goal of this project is to develop methods which integrate genome, transcriptome and trait data to identify and validate impactful non-coding variants. Funding for Stanford is split between 3 PIs. (On NCX to 2019)

1R33HL120757 (Quertermous, Thomas) 03/05/2014-02/28/2019 0.60 calendar
National Institutes of Health
Identification of Causal Coronary Heart Disease Variation in Smooth Muscle Cells
Major Goals: The main goal of this project is to apply genome, transcriptome and epigenome sequencing to identify causal regulatory variants underlying coronary heart disease-associated variation.
Role: Co-investigator. I assisted with grant writing and this funding supports the work of Boxiang Liu (a graduate student in my lab) who is leading work on analyzing eQTLs in coronary artery smooth muscle cells between our lab and the Quertermous Lab.

Grant 02/01/2016-01/31/2019 0.00 calendar*
Glenn Center for Biology of Aging Research Stanford University
Genetics of Gene Expression in an Elderly, Longitudinal Cohort
Major Goals: Main goals are longitudinal transcriptome profiling of elderly individuals; and identification of gene regulatory changes in advanced age.
*PI effort may not be billed to Glenn Center grant.

1R01MH101814 (Bustamante, Carlos D.) 08/01/2013-06/30/2017 0.84 calendar
National Institutes of Health University of Geneva
Methods for High-Resolution Analysis of Genetic Effects on Gene Expression
Major Goals: This project is for the second phase of statistical analyses as part of GTEx.
Role: Co-investigator. I helped write this grant. I co-led multiple aspects of the second phase of GTEx that culminated in a set of papers in the Oct 12th, 2017 issue of Nature.

5U01HG007436 (Bustamante, Carlos D.) 09/23/2013-07/31/2017 0.60 calendar
National Institutes of Health Clinically Relevant Genome Variation Database
Major Goals: The main goal of this project is to create a unified, public and freely available database of genetic alterations relevant to clinical care. Our ultimate goal is to empower clinicians, genetic counselors, and patients to make informed decisions based on DNA testing.
Role: Co-investigator. I helped write a small part of this grant. My lab focused on machine learning approaches for causal variants in disease.

1U01HG007593-03 (Li, Jin Billy) 04/22/2014-03/31/2017 1.80 calendar

National Institutes of Health
High Resolution Allele Specific Expression Assays
Role: Co-investigator. I helped write this grant. My lab is co-leading various aspects of eGTEx data production analysis and coordinated a marker paper in Nature Genetics in 2017.

54341548 (Montgomery, Stephen) 09/11/2014 - 05/31/2016 0.60 calendar
Prime Sponsor: National Institutes of Health
University of California, San Diego
Single-cell Sequencing and in situ Mapping of RNA Transcripts in Human Brains

N/A (Montgomery, Stephen) 10/01/2012 - 09/30/2015 1.44 calendar
Edward Mallinckrodt, Jr. Foundation
Expression Response to Environment in Coronary Artery Smooth Muscle Cells

The Fred Hutchinson Cancer Research Center 03/01/2015 – 08/31/2015 0.12 calendar
Pilot RNA-seq Study Among Long Life Study Participants of the WHI