Stanford

Stephen B. Montgomery

Professor of Pathology, of Genetics and of Biomedical Data Science

CONTACT INFORMATION

- **Alternate Contact**

  Char Armitage - Administrative Assistant

  Email carmitag@stanford.edu

Bio

ACADEMIC APPOINTMENTS

- Professor, Pathology
- Professor, Genetics
- Professor, Biomedical Data Science
- Member, Bio-X
- Member, Cardiovascular Institute
- Member, Wu Tsai Human Performance Alliance
- 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/
- GREGoR Stanford Site: https://gregor.stanford.edu/

Research & Scholarship

CURRENT RESEARCH AND SCHOLARLY INTERESTS

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

Teaching

COURSES

2023-24

- Informatics in Industry: BIOMEDIN 206 (Spr)

2022-23

- Informatics in Industry: BIOMEDIN 206 (Spr)

2020-21

- Foundations in Experimental Biology: BIOS 200 (Aut)

STANFORD ADVISEES

Doctoral Dissertation Reader (AC)

Michael Hayes, Jodie Lunger, Roshni Patel, Jack Shanahan

Postdoctoral Faculty Sponsor

Iman Jaljuli, Nick Lashinsky, Andrew Marderstein, Evin Padhi, Yilin Xie

Doctoral Dissertation Advisor (AC)

Tiffany Eulalio, Page Goddard, Emily Greenwald, Tanner Jensen, Julie Lake, Kate Lawrence, Jarod Rutledge, Rachel Ungar

Master's Program Advisor

Francesca Bottazzini, Zachary Cadiz, Caroline Van

Doctoral Dissertation Co-Advisor (AC)

Kameron Rodrigues

Doctoral (Program)

Ben Ehlert, Samson Mataraso, Esther Robb, Min Sun, Ben Viggiano, Christine Yeh, Juan Manuel Zambrano Chaves

Postdoctoral Research Mentor

Daniel Nachun

GRADUATE AND FELLOWSHIP PROGRAM AFFILIATIONS

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

Publications

PUBLICATIONS

- **Integrative analyses highlight functional regulatory variants associated with neuropsychiatric diseases.** *Nature genetics*
  Guo, M. G., Reynolds, D. L., Ang, C. E., Liu, Y., Zhao, Y., Donohue, L. K., Siprashvili, Z., Yang, X., Yoo, Y., Mondal, S., Hong, A., Kain, J., Meservey, et al
  2023
- **The functional impact of rare variation across the regulatory cascade.** *Cell genomics*
  Li, T., Ferraro, N., Strober, B. J., Aguet, F., Kasela, S., Arvanitis, M., Ni, B., Wiel, L., Hershberg, E., Ardlie, K., Arking, D. E., Beer, R. L., Brody, et al
  2023; 3 (10): 100401
- **Integrated single-cell multiome analysis reveals muscle fiber-type gene regulatory circuitry modulated by endurance exercise.** *bioRxiv : the preprint server for biology*

Rubenstein, A. B., Smith, G. R., Zhang, Z., Chen, X., Chambers, T. L., Ruf-Zamojski, F., Mendelev, N., Cheng, W. S., Zamojski, M., Amper, M. A., Nair, V. D., Marderstein, A. R., Montgomery, et al

2023

- **Author Correction: Africa-specific human genetic variation near CHD1L associates with HIV-1 load.** *Nature*

  McLaren, P. J., Porreca, I., Iaconis, G., Mok, H. P., Mukhopadhyay, S., Karakoc, E., Cristinelli, S., Pomilla, C., Bartha, I., Thorball, C. W., Tough, R. H., Angelino, P., Kiar, et al

  2023

- **Beyond the exome: What's next in diagnostic testing for Mendelian conditions.** *American journal of human genetics*

  Wojcik, M. H., Reuter, C. M., Marwaha, S., Mahmoud, M., Duyzend, M. H., Barseghyan, H., Yuan, B., Boone, P. M., Groopman, E. E., Délot, E. C., Jain, D., Sanchis-Juan, A., Starita, et al

  2023; 110 (8): 1229-1248

- **Africa-specific human genetic variation near CHD1L associates with HIV-1 load.** *Nature*

  McLaren, P. J., Porreca, I., Iaconis, G., Mok, H. P., Mukhopadhyay, S., Karakoc, E., Cristinelli, S., Pomilla, C., Bartha, I., Thorball, C. W., Tough, R. H., Angelino, P., Kiar, et al

  2023

- **Molecular quantitative trait loci** *NATURE REVIEWS METHODS PRIMERS*

  Aguet, F., Alasoo, K., Li, Y., Battle, A., Im, H., Montgomery, S. B., Lappalainen, T.

  2023; 3 (1)

- **Beyond the exome: what's next in diagnostic testing for Mendelian conditions.** *ArXiv*

  Wojcik, M. H., Reuter, C. M., Marwaha, S., Mahmoud, M., Duyzend, M. H., Barseghyan, H., Yuan, B., Boone, P. M., Groopman, E. E., Délot, E. C., Jain, D., Sanchis-Juan, A., Starita, et al

  2023

- **The mitochondrial multi-omic response to exercise training across tissues.** *bioRxiv : the preprint server for biology*

  Amar, D., Gay, N. R., Jimenez-Morales, D., Beltran, P. M., Ramaker, M. E., Raja, A. N., Zhao, B., Sun, Y., Marwaha, S., Gaul, D., Hershman, S. G., Xia, A., Lanza, et al

  2023

- **Multiomic identification of key transcriptional regulatory programs during endurance exercise training.** *bioRxiv : the preprint server for biology*

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

  2023

- **RNAget: an API to securely retrieve RNA quantifications.** *Bioinformatics (Oxford, England)*

  Upchurch, S., Palumbo, E., Adams, J., Bujold, D., Bourque, G., Nedzel, J., Graham, K., Kagda, M. S., Assis, P., Hitz, B., Righi, E., Guigo, R., Wold, et al

  2023; 39 (4)

- **Methylation differences in Alzheimer's disease neuropathologic change in the aged human brain.** *Acta neuropathologica communications*

  Lang, A. L., Eulalio, T., Fox, E., Yakabi, K., Bukhari, S. A., Kawas, C. H., Corrada, M. M., Montgomery, S. B., Heppner, F. L., Capper, D., Nachun, D., Montine, T. J.

  2022; 10 (1): 174

- **Deep learning-assisted genome-wide characterization of massively parallel reporter assays.** *Nucleic acids research*

  Lu, F., Sossin, A., Abell, N., Montgomery, S. B., He, Z.

  2022

- **RNA editing underlies genetic risk of common inflammatory diseases.** *Nature*

  Li, Q., Gloudemans, M. J., Geisinger, J. M., Fan, B., Aguet, F., Sun, T., Ramaswami, G., Li, Y. I., Ma, J. B., Pritchard, J. K., Montgomery, S. B., Li, J. B.

  2022

- **Temporal dynamics of the multi-omic response to endurance exercise training across tissues**

  Gay, N. R., Beltran, P., Amar, D., Montgomery, S. B., Carr, S. A., Motrpac Study Grp

  ELSEVIER.2022: S31

- **Integration of rare expression outlier-associated variants improves polygenic risk prediction.** *American journal of human genetics*

  Smail, C., Ferraro, N. M., Hui, Q., Durrant, M. G., Aguirre, M., Tanigawa, Y., Keever-Keigher, M. R., Rao, A. S., Justesen, J. M., Li, X., Gloudemans, M. J., Assimes, T. L., Kooperberg, et al

2022

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

- **Single-cell epigenomic analyses implicate candidate causal variants at inherited risk loci for Alzheimer's and Parkinson's diseases.** *Nature genetics*
  Corces, M. R., Shcherbina, A., Kundu, S., Gloudemans, M. J., Fresard, L., Granja, J. M., Louie, B. H., Eulalio, T., Shams, S., Bagdatli, S. T., Mumbach, M. R., Liu, B., Montine, et al
  2020

- **The GTEx Consortium atlas of genetic regulatory effects across human tissues** *SCIENCE*
  Aguet, F., Barbeira, A. N., Bonazzola, R., Brown, A., Castel, S. E., Jo, B., Kasela, S., Kim-Hellmuth, S., Liang, Y., Parsana, P., Flynn, E., Fresard, L., Gamazon, et al
  2020; 369 (6509): 1318-+

- **Molecular Transducers of Physical Activity Consortium (MoTrPAC): Mapping the Dynamic Responses to Exercise.** *Cell*
  Sanford, J. A., Nogiec, C. D., Lindholm, M. E., Adkins, J. N., Amar, D., Dasari, S., Drugan, J. K., Fernandez, F. M., Radom-Aizik, S., Schenk, S., Snyder, M. P., Tracy, R. P., Vanderboom, et al
  2020; 181 (7): 1464–74

- **Discovery and quality analysis of a comprehensive set of structural variants and short tandem repeats.** *Nature communications*
  Jakubosky, D., Smith, E. N., D'Antonio, M., Jan Bonder, M., Young Greenwald, W. W., D'Antonio-Chronowska, A., Matsui, H., i2QTL Consortium, Stegle, O., Montgomery, S. B., DeBoever, C., Frazer, K. A., Bonder, M. J., et al
  2020; 11 (1): 2928

- **Properties of structural variants and short tandem repeats associated with gene expression and complex traits.** *Nature communications*
  Jakubosky, D., D'Antonio, M., Bonder, M. J., Smail, C., Donovan, M. K., Young Greenwald, W. W., Matsui, H., i2QTL Consortium, D'Antonio-Chronowska, A., Stegle, O., Smith, E. N., Montgomery, S. B., DeBoever, C., et al
  2020; 11 (1): 2927

- **Transcriptional and Position Effect Contributions to rAAV-Mediated Gene Targeting**
  Spector, L. P., Tiffany, M., Ferraro, N. M., Abell, N. S., Montgomery, S. B., Kay, M. A.
  CELL PRESS.2020: 290

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

- **Evaluating the genomic parameters governing rAAV-mediated homologous recombination.** *Molecular therapy : the journal of the American Society of Gene Therapy*
  Spector, L. P., Tiffany, M. n., Ferraro, N. M., Abell, N. S., Montgomery, S. B., Kay, M. A.
  2020

- **The impact of sex on gene expression across human tissues.** *Science (New York, N.Y.)*
  Oliva, M. n., Muñoz-Aguirre, M. n., Kim-Hellmuth, S. n., Wucher, V. n., Gewirtz, A. D., Cotter, D. J., Parsana, P. n., Kasela, S. n., Balliu, B. n., Viñuela, A. n., Castel, S. E., Mohammadi, P. n., Aguet, et al
  2020; 369 (6509)

- **Impact of admixture and ancestry on eQTL analysis and GWAS colocalization in GTEx.** *Genome biology*
  Gay, N. R., Gloudemans, M. n., Antonio, M. L., Abell, N. S., Balliu, B. n., Park, Y. n., Martin, A. R., Musharoff, S. n., Rao, A. S., Aguet, F. n., Barbeira, A. N., Bonazzola, R. n., Hormozdiari, et al

2020; 21 (1): 233

- **Transcriptomic signatures across human tissues identify functional rare genetic variation.** *Science (New York, N.Y.)*
  Ferraro, N. M., Strober, B. J., Einson, J. n., Abell, N. S., Aguet, F. n., Barbeira, A. N., Brandt, M. n., Bucan, M. n., Castel, S. E., Davis, J. R., Greenwald, E. n., Hess, G. T., Hilliard, et al
  2020; 369 (6509)

- **FAM13A affects body fat distribution and adipocyte function.** *Nature communications*
  Fathzadeh, M. n., Li, J. n., Rao, A. n., Cook, N. n., Chennamsetty, I. n., Seldin, M. n., Zhou, X. n., Sangwung, P. n., Gloudemans, M. J., Keller, M. n., Attie, A. n., Yang, J. n., Wabitsch, et al
  2020; 11 (1): 1465

- **A Bioinformatic Analysis of Integrative Mobile Genetic Elements Highlights Their Role in Bacterial Adaptation.** *Cell host & microbe*
  Durrant, M. G., Li, M. M., Siranosian, B. A., Montgomery, S. B., Bhatt, A. S.
  2019

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

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

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

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

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

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

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

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

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

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

- **Proficiency Testing of Standardized Samples Shows Very High Interlaboratory Agreement for Clinical Next-Generation Sequencing-Based Oncology Assays** *ARCHIVES OF PATHOLOGY & LABORATORY MEDICINE*

Merker, J. D., Devereaux, K., Iafrate, A., Kamel-Reid, S., Kim, A. S., Moncur, J. T., Montgomery, S. B., Nagarajan, R., Portier, B. P., Routbort, M. J., Smail, C., Surrey, L. F., Vasalos, et al

2019; 143 (4): 463–71

- **A toolkit for genetics providers in follow-up of patients with non-diagnostic exome sequencing** *JOURNAL OF GENETIC COUNSELING*

  Zastrow, D. B., Kohler, J. N., Bonner, D., Reuter, C. M., Fernandez, L., Grove, M. E., Fisk, D. G., Yang, Y., Eng, C. M., Ward, P. A., Bick, D., Worthey, E. A., Fisher, et al

  2019; 28 (2): 213–28

- **Identification of 22 novel loci associated withurinary biomarkers of albumin, sodium, andpotassium excretion.** *Kidney international*

  Zanetti, D., Rao, A., Gustafsson, S., Assimes, T. L., Montgomery, S. B., Ingelsson, E.

  2019

- **Abundant associations with gene expression complicate GWAS follow-up.** *Nature genetics*

  Liu, B. n., Gloudemans, M. J., Rao, A. S., Ingelsson, E. n., Montgomery, S. B.

  2019; 51 (5): 768–69

- **SEX DIFFERENCES AT THE MOLECULAR LEVEL: LESSONS FROM THE HUMAN TRANSCRIPTOME**

  Stranger, B., Oliva, M., Gamazon, E., Reverter, F., Wucher, V., Balliu, B., Dumitrascu, B., Parsana, P., Payne, A., Jo, B., Montgomery, S., Battle, A., Ardlie, et al

  ELSEVIER.2019: 1034

- **A toolkit for genetics providers in follow-up of patients with non-diagnostic exome sequencing.** *Journal of genetic counseling*

  Zastrow, D. B., Kohler, J. N., Bonner, D. n., Reuter, C. M., Fernandez, L. n., Grove, M. E., Fisk, D. G., Yang, Y. n., Eng, C. M., Ward, P. A., Bick, D. n., Worthey, E. A., Fisher, et al

  2019; 28 (2): 213–28

- **Genetic analyses of human fetal retinal pigment epithelium gene expression suggest ocular disease mechanisms.** *Communications biology*

  Liu, B., Calton, M. A., Abell, N. S., Benchorin, G., Gloudemans, M. J., Chen, M., Hu, J., Li, X., Balliu, B., Bok, D., Montgomery, S. B., Vollrath, D.

  2019; 2: 186

- **Pathologic gene network rewiring implicates PPP1R3A as a central regulator in pressure overload heart failure.** *Nature communications*

  Cordero, P., Parikh, V. N., Chin, E. T., Erbilgin, A., Gloudemans, M. J., Shang, C., Huang, Y., Chang, A. C., Smith, K. S., Dewey, F., Zaleta, K., Morley, M., Brandimarto, et al

  2019; 10 (1): 2760

- **Identification of rare-disease genes using blood transcriptome sequencing and large control cohorts.** *Nature medicine*

  Frésard, L. n., Smail, C. n., Ferraro, N. M., Teran, N. A., Li, X. n., Smith, K. S., Bonner, D. n., Kernohan, K. D., Marwaha, S. n., Zappala, Z. n., Balliu, B. n., Davis, J. R., Liu, et al

  2019

- **Diagnosing rare diseases after the exome.** *Cold Spring Harbor molecular case studies*

  Fresard, L., Montgomery, S. B.

  2018; 4 (6)

- **Proficiency Testing of Standardized Samples Shows Very High Interlaboratory Agreement for Clinical Next-Generation Sequencing-Based Oncology Assays.** *Archives of pathology & laboratory medicine*

  Merker, J. D., Devereaux, K., Iafrate, A. J., Kamel-Reid, S., Kim, A. S., Moncur, J. T., Montgomery, S. B., Nagarajan, R., Portier, B. P., Routbort, M. J., Smail, C., Surrey, L. F., Vasalos, et al

  2018

- **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.

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