Stanford

Manuel Rivas

Assistant Professor of Biomedical Data Science

NIH Biosketch available Online

Bio

ACADEMIC APPOINTMENTS

- Assistant Professor, Biomedical Data Science
- Member, Bio-X

HONORS AND AWARDS

- Clarendon Scholar, University of Oxford (2010-2015)
- Osler Award, University of Oxford (2010-2015)
- Gates Millenium Scholar, Bill & Melinda Gates Foundation (2004-2008)

PROFESSIONAL EDUCATION

- DPhil, University of Oxford , Clinical Medicine (2015)
- B.S., Massachusetts Institute of Technology , Mathematics (2008)

LINKS

- My Lab Site: http://med.stanford.edu/rivaslab

Teaching

COURSES

2020-21

- Analytics Accelerator: BIODS 217, CME 217 (Aut, Win)
- Cloud Computing for Biology and Healthcare: GENE 222 (Spr)
- Topics in Biomedical Data Science: Large-scale inference: BIODS 215 (Win)
- Workshop in Biostatistics: BIODS 260B, STATS 260B (Win)

2019-20

- Topics in Biomedical Data Science: Large-scale inference: BIODS 215 (Win)
- Workshop in Biostatistics: BIODS 260B, STATS 260B (Win)

2018-19

- Workshop in Biostatistics: BIODS 260C, STATS 260C (Spr)

2017-18

- Topics in Biomedical Data Science: Large-scale inference: BIODS 215 (Win)

- Workshop in Biostatistics: BIODS 260C, STATS 260C (Spr)

STANFORD ADVISEES

Doctoral Dissertation Reader (AC)

E Flynn, Nicole Gay, Rosa Ma, Greg McInnes, Kelley Paskov

Postdoctoral Faculty Sponsor

Yongchan Kwon, Alice Popejoy

Doctoral Dissertation Advisor (AC)

Yosuke Tanigawa, Guhan Venkataraman

Doctoral Dissertation Co-Advisor (AC)

Ruilin Li

Publications

PUBLICATIONS

- **Survival Analysis on Rare Events Using Group-Regularized Multi-Response Cox Regression.** *Bioinformatics (Oxford, England)*
  Li, R., Tanigawa, Y., Justesen, J. M., Taylor, J., Hastie, T., Tibshirani, R., Rivas, M. A.
  2021

- **Polygenic risk modeling with latent trait-related genetic components.** *European journal of human genetics : EJHG*
  Aguirre, M., Tanigawa, Y., Venkataraman, G. R., Tibshirani, R., Hastie, T., Rivas, M. A.
  2021

- **Genetics of 35 blood and urine biomarkers in the UK Biobank.** *Nature genetics*
  Sinnott-Armstrong, N., Tanigawa, Y., Amar, D., Mars, N., Benner, C., Aguirre, M., Venkataraman, G. R., Wainberg, M., Ollila, H. M., Kiiskinen, T., Havulinna, A. S., Pirruccello, J. P., Qian, et al
  2021

- **Graphical analysis for phenome-wide causal discovery in genotyped population-scale biobanks.** *Nature communications*
  Amar, D., Sinnott-Armstrong, N., Ashley, E. A., Rivas, M. A.
  2021; 12 (1): 350

- **Sex-specific genetic effects across biomarkers.** *European journal of human genetics : EJHG*
  Flynn, E., Tanigawa, Y., Rodriguez, F., Altman, R. B., Sinnott-Armstrong, N., Rivas, M. A.
  2020

- **Rare protein-altering variants in ANGPTL7 lower intraocular pressure and protect against glaucoma** *PLOS GENETICS*
  Tanigawa, Y., Wainberg, M., Karjalainen, J., Kiiskinen, T., Venkataraman, G., Lemmela, S., Turunen, J. A., Graham, R. R., Havulinna, A. S., Perola, M., Palotie, A., Gen, F., Daly, et al
  2020; 16 (5)

- **A fast and scalable framework for large-scale and ultrahigh-dimensional sparse regression with application to the UK Biobank.** *PLoS genetics*
  Qian, J. n., Tanigawa, Y. n., Du, W. n., Aguirre, M. n., Chang, C. n., Tibshirani, R. n., Rivas, M. A., Hastie, T. n.
  2020; 16 (10): e1009141

- **Assessing Digital Phenotyping to Enhance Genetic Studies of Human Diseases.** *American journal of human genetics*
  DeBoever, C. n., Tanigawa, Y. n., Aguirre, M. n., McInnes, G. n., Lavertu, A. n., Rivas, M. A.
  2020

- **Fast Lasso method for large-scale and ultrahigh-dimensional Cox model with applications to UK Biobank.** *Biostatistics (Oxford, England)*
  Li, R. n., Chang, C. n., Justesen, J. M., Tanigawa, Y. n., Qiang, J. n., Hastie, T. n., Rivas, M. A., Tibshirani, R. n.
  2020

- **Homogeneity in the association of body mass index with type 2 diabetes across the UK Biobank: A Mendelian randomization study.** *PLoS medicine*
  Wainberg, M., Mahajan, A., Kundaje, A., McCarthy, M. I., Ingelsson, E., Sinnott-Armstrong, N., Rivas, M. A.
  2019; 16 (12): e1002982

- **Rare and common variant discovery in complex disease: the IBD case study.** *Human molecular genetics*
  Venkataraman, G. R., Rivas, M. A.
  2019

- **Phenome-wide Burden of Copy-Number Variation in the UK Biobank.** *American journal of human genetics*
  Aguirre, M., Rivas, M. A., Priest, J.
  2019

- **Global Biobank Engine: enabling genotype-phenotype browsing for biobank summary statistics** *BIOINFORMATICS*
  McInnes, G., Tanigawa, Y., DeBoever, C., Lavertu, A., Olivieri, J., Aguirre, M., Rivas, M. A.
  2019; 35 (14): 2495–97

- **Opportunities and challenges for transcriptome-wide association studies** *NATURE GENETICS*
  Wainberg, M., Sinnott-Armstrong, N., Mancuso, N., Barbeira, A. N., Knowles, D. A., Golan, D., Ermel, R., Ruusalepp, A., Quertermous, T., Hao, K., Bjorkegren, J. M., Im, H., Pasaniuc, et al
  2019; 51 (4): 592–99

- **Significant shared heritability underlies suicide attempt and clinically predicted probability of attempting suicide.** *Molecular psychiatry*
  Ruderfer, D. M., Walsh, C. G., Aguirre, M. W., Tanigawa, Y., Ribeiro, J. D., Franklin, J. C., Rivas, M. A.
  2019

- **Components of genetic associations across 2,138 phenotypes in the UK Biobank highlight adipocyte biology.** *Nature communications*
  Tanigawa, Y. n., Li, J. n., Justesen, J. M., Horn, H. n., Aguirre, M. n., DeBoever, C. n., Chang, C. n., Narasimhan, B. n., Lage, K. n., Hastie, T. n., Park, C. Y., Bejerano, G. n., Ingelsson, et al
  2019; 10 (1): 4064

- **Opportunities and challenges for transcriptome-wide association studies.** *Nature genetics*
  Wainberg, M. n., Sinnott-Armstrong, N. n., Mancuso, N. n., Barbeira, A. N., Knowles, D. A., Golan, D. n., Ermel, R. n., Ruusalepp, A. n., Quertermous, T. n., Hao, K. n., Björkegren, J. L., Im, H. K., Pasaniuc, et al
  2019; 51 (4): 592–99

- **Global Biobank Engine: enabling genotype-phenotype browsing for biobank summary statistics.** *Bioinformatics (Oxford, England)*
  McInnes, G., Tanigawa, Y., DeBoever, C., Lavertu, A., Olivieri, J. E., Aguirre, M., Rivas, M. A.
  2018

- **Medical relevance of protein-truncating variants across 337,205 individuals in the UK Biobank study** *NATURE COMMUNICATIONS*
  DeBoever, C., Tanigawa, Y., Lindholm, M. E., McInnes, G., Lavertu, A., Ingelsson, E., Chang, C., Ashley, E. A., Bustamante, C. D., Daly, M. J., Rivas, M. A.
  2018; 9: 1612

- **Insights into the genetic epidemiology of Crohn's and rare diseases in the Ashkenazi Jewish population.** *PLoS genetics*
  Rivas, M. A., Avila, B. E., Koskela, J., Huang, H., Stevens, C., Pirinen, M., Haritunians, T., Neale, B. M., Kurki, M., Ganna, A., Graham, D., Glaser, B., Peter, et al
  2018; 14 (5): e1007329

- **DeepTag: inferring diagnoses from veterinary clinical notes.** *NPJ digital medicine*
  Nie, A. n., Zehnder, A. n., Page, R. L., Zhang, Y. n., Pineda, A. L., Rivas, M. A., Bustamante, C. D., Zou, J. n.
  2018; 1: 60

- **A protein-truncating R179X variant in RNF186 confers protection against ulcerative colitis** *NATURE COMMUNICATIONS*
  Rivas, M. A., Graham, D., Sulem, P., Stevens, C., Desch, A. N., Goyette, P., Gudbjartsson, D., Jonsdottir, I., Thorsteinsdottir, U., Degenhardt, F., Mucha, S., Kurki, M. I., Li, et al
  2016; 7

- **Discovery of rare variants for complex phenotypes** *HUMAN GENETICS*
  Kosmicki, J. A., Churchhouse, C. L., Rivas, M. A., Neale, B. M.
  2016; 135 (6): 625-634

- **Assessing allele-specific expression across multiple tissues from RNA-seq read data** *BIOINFORMATICS*
  Pirinen, M., Lappalainen, T., Zaitlen, N. A., Dermitzakis, E. T., Donnelly, P., McCarthy, M. I., Rivas, M. A.
  2015; 31 (15): 2497-2504

- **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

- **The Power of Gene-Based Rare Variant Methods to Detect Disease-Associated Variation and Test Hypotheses About Complex Disease** *PLOS GENETICS*
  Moutsianas, L., Agarwala, V., Fuchsberger, C., Flannick, J., Rivas, M. A., Gaulton, K. J., Albers, P. K., McVean, G., Boehnke, M., Altshuler, D., McCarthy, M. I.
  2015; 11 (4)

- **Choice of transcripts and software has a large effect on variant annotation** *GENOME MEDICINE*
  McCarthy, D. J., Humburg, P., Kanapin, A., Rivas, M. A., Gaulton, K., Cazier, J., Donnelly, P.
  2014; 6

- **Assessing association between protein truncating variants and quantitative traits** *BIOINFORMATICS*
  Rivas, M. A., Pirinen, M., Neville, M. J., Gaulton, K. J., Moutsianas, L., Lindgren, C. M., Karpe, F., McCarthy, M. I., Donnelly, P.
  2013; 29 (19): 2419-2426

- **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

- **Deep Resequencing of GWAS Loci Identifies Rare Variants in CARD9, IL23R and RNF186 That Are Associated with Ulcerative Colitis** *PLOS GENETICS*
  Beaudoin, M., Goyette, P., Boucher, G., Lo, K. S., Rivas, M. A., Stevens, C., Alikashani, A., Ladouceur, M., Ellinghaus, D., Torkvist, L., Goel, G., Lagace, C., Annese, et al
  2013; 9 (9)

- **A Flexible Approach for the Analysis of Rare Variants Allowing for a Mixture of Effects on Binary or Quantitative Traits** *PLOS GENETICS*
  Clarke, G. M., Rivas, M. A., Morris, A. P.
  2013; 9 (8)

- **Deep resequencing of GWAS loci identifies independent rare variants associated with inflammatory bowel disease** *NATURE GENETICS*
  Rivas, M. A., Beaudoin, M., Gardet, A., Stevens, C., Sharma, Y., Zhang, C. K., Boucher, G., Ripke, S., Ellinghaus, D., Burtt, N., Fennell, T., Kirby, A., Latiano, et al
  2011; 43 (11): 1066-U50

- **A framework for variation discovery and genotyping using next-generation DNA sequencing data** *NATURE GENETICS*
  DePristo, M. A., Banks, E., Poplin, R., Garimella, K. V., Maguire, J. R., Hartl, C., Philippakis, A. A., del Angel, G., Rivas, M. A., Hanna, M., McKenna, A., Fennell, T. J., Kernytsky, et al
  2011; 43 (5): 491-?

- **Testing for an Unusual Distribution of Rare Variants** *PLOS GENETICS*
  Neale, B. M., Rivas, M. A., Voight, B. F., Altshuler, D., Devlin, B., Orho-Melander, M., Kathiresan, S., Purcell, S. M., Roeder, K., Daly, M. J.
  2011; 7 (3)

- **A regulatory variant at 3q21.1 confers an increased pleiotropic risk for hyperglycemia and altered bone mineral density.** *Cell metabolism*
  Sinnott-Armstrong, N., Sousa, I. S., Laber, S., Rendina-Ruedy, E., Nitter Dankel, S. E., Ferreira, T., Mellgren, G., Karasik, D., Rivas, M., Pritchard, J., Guntur, A. R., Cox, R. D., Lindgren, et al
  2021

- **Sleep apnoea is a risk factor for severe COVID-19.** *BMJ open respiratory research*
  Strausz, S., Kiiskinen, T., Broberg, M., Ruotsalainen, S., Koskela, J., Bachour, A., FinnGen, Palotie, A., Palotie, T., Ripatti, S., Ollila, H. M., Palotie, A., Daly, M., et al
  2021; 8 (1)

- **Significant shared heritability underlies suicide attempt and clinically predicted probability of attempting suicide** *MOLECULAR PSYCHIATRY*
  Ruderfer, D. M., Walsh, C. G., Aguirre, M. W., Tanigawa, Y., Ribeiro, J. D., Franklin, J. C., Rivas, M. A.

2020; 25 (10): 2422–30

- **Race, socioeconomic deprivation, and hospitalization for COVID-19 in English participants of a national biobank.** *International journal for equity in health*
  Patel, A. P., Paranjpe, M. D., Kathiresan, N. P., Rivas, M. A., Khera, A. V.
  2020; 19 (1): 114

- **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

- **Rare protein-altering variants in ANGPTL7 lower intraocular pressure and protect against glaucoma.** *PLoS genetics*
  Tanigawa, Y., Wainberg, M., Karjalainen, J., Kiiskinen, T., Venkataraman, G., Lemmela, S., Turunen, J. A., Graham, R. R., Havulinna, A. S., Perola, M., Palotie, A., FinnGen, Daly, M. J., et al
  2020; 16 (5): e1008682

- **Whole exome sequencing analyses reveal gene-microbiota interactions in the context of IBD.** *Gut*
  Hu, S. n., Vich Vila, A. n., Gacesa, R. n., Collij, V. n., Stevens, C. n., Fu, J. M., Wong, I. n., Talkowski, M. E., Rivas, M. A., Imhann, F. n., Bolte, L. n., van Dullemen, H. n., Dijkstra, et al
  2020

- **A phenome-wide association study of 26 mendelian genes reveals phenotypic expressivity of common and rare variants within the general population.** *PLoS genetics*
  Tcheandjieu, C. n., Aguirre, M. n., Gustafsson, S. n., Saha, P. n., Potiny, P. n., Haendel, M. n., Ingelsson, E. n., Rivas, M. A., Priest, J. R.
  2020; 16 (11): e1008802

- **FasTag: Automatic text classification of unstructured medical narratives.** *PloS one*
  Venkataraman, G. R., Pineda, A. L., Bear Don't Walk Iv, O. J., Zehnder, A. M., Ayyar, S., Page, R. L., Bustamante, C. D., Rivas, M. A.
  2020; 15 (6): e0234647

- **Cardiac Imaging of Aortic Valve Area from 34,287 UK Biobank Participants Reveal Novel Genetic Associations and Shared Genetic Comorbidity with Multiple Disease Phenotypes.** *Circulation. Genomic and precision medicine*
  Córdova-Palomera, A. n., Tcheandjieu, C. n., Fries, J. n., Varma, P. n., Chen, V. S., Fiterau, M. n., Xiao, K. n., Tejeda, H. n., Keavney, B. n., Cordell, H. J., Tanigawa, Y. n., Venkataraman, G. n., Rivas, et al
  2020

- **The role of polygenic risk and susceptibility genes in breast cancer over the course of life.** *Nature communications*
  Mars, N., Widen, E., Kerminen, S., Meretoja, T., Pirinen, M., Della Briotta Parolo, P., Palta, P., FinnGen, Palotie, A., Kaprio, J., Joensuu, H., Daly, M., Ripatti, S., et al
  2020; 11 (1): 6383

- **Genetic architecture of human plasma lipidome and its link to cardiovascular disease.** *Nature communications*
  Tabassum, R., Ramo, J. T., Ripatti, P., Koskela, J. T., Kurki, M., Karjalainen, J., Palta, P., Hassan, S., Nunez-Fontarnau, J., Kiiskinen, T. T., Soderlund, S., Matikainen, N., Gerl, et al
  2019; 10 (1): 4329

- **Protein-coding variants implicate novel genes related to lipid homeostasis contributing to body-fat distribution** *NATURE GENETICS*
  Justice, A. E., Karaderi, T., Highland, H. M., Young, K. L., Graff, M., Lu, Y., Turcot, V., Auer, P. L., Fine, R. S., Guo, X., Schurmann, C., Lempradl, A., Marouli, et al
  2019; 51 (3): 452-+

- **Association of Genetic Variants in NUDT15 With Thiopurine-Induced Myelosuppression in Patients With Inflammatory Bowel Disease.** *JAMA*
  Walker, G. J., Harrison, J. W., Heap, G. A., Voskuil, M. D., Andersen, V., Anderson, C. A., Ananthakrishnan, A. N., Barrett, J. C., Beaugerie, L., Bewshea, C. M., Cole, A. T., Cummings, F. R., Daly, et al
  2019; 321 (8): 773–85

- **Association of Genetic Variants in NUDT15 With Thiopurine-Induced Myelosuppression in Patients With Inflammatory Bowel Disease** *JAMA-JOURNAL OF THE AMERICAN MEDICAL ASSOCIATION*
  Walker, G. J., Harrison, J. W., Heap, G. A., Voskuil, M. D., Andersen, V., Anderson, C. A., Ananthakrishnan, A. N., Barrett, J. C., Beaugerie, L., Bewshea, C. M., Cole, A. T., Cummings, F. R., Daly, et al
  2019; 321 (8): 773–85

- **Protein-coding variants implicate novel genes related to lipid homeostasis contributing to body-fat distribution.** *Nature genetics*
  Justice, A. E., Karaderi, T., Highland, H. M., Young, K. L., Graff, M., Lu, Y., Turcot, V., Auer, P. L., Fine, R. S., Guo, X., Schurmann, C., Lempradl, A., Marouli, et al
  2019

- **DeepTag: inferring diagnoses from veterinary clinical notes** *NPJ DIGITAL MEDICINE*
  Nie, A., Zehnder, A., Page, R. L., Zhang, Y., Pineda, A., Rivas, M. A., Bustamante, C. D., Zou, J.
  2018; 1

- **Base-specific mutational intolerance near splice sites clarifies the role of nonessential splice nucleotides** *GENOME RESEARCH*
  Zhang, S., Samocha, K. E., Rivas, M. A., Karczewski, K. J., Daly, E., Schmandt, B., Neale, B. M., MacArthur, D. G., Daly, M. J.
  2018; 28 (7): 968–74

- **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

- **Quantifying the Impact of Rare and Ultra-rare Coding Variation across the Phenotypic Spectrum.** *American journal of human genetics*
  Ganna, A., Satterstrom, F. K., Zekavat, S. M., Das, I., Kurki, M. I., Churchhouse, C., Alfoldi, J., Martin, A. R., Havulinna, A. S., Byrnes, A., Thompson, W. K., Nielsen, P. R., Karczewski, et al
  2018

- **Genetic variants in cellular transport do not affect mesalamine response in ulcerative colitis** *PLOS ONE*
  Moran, C. J., Huang, H., Rivas, M., Kaplan, J. L., Daly, M. J., Winter, H. S.
  2018; 13 (3): e0192806

- **Sequence data and association statistics from 12,940 type 2 diabetes cases and controls (vol 4, 170179, 2017)** *SCIENTIFIC DATA*
  Flannick, J., Fuchsberger, C., Mahajan, A., Teslovich, T. M., Agarwala, V., Gaulton, K. J., Caulkins, L., Koesterer, R., Ma, C., Moutsianas, L., McCarthy, D. J., Rivas, M. A., Perry, et al
  2018; 5: 180002

- **Evaluating the contribution of rare variants to type 2 diabetes and related traits using pedigrees** *PROCEEDINGS OF THE NATIONAL ACADEMY OF SCIENCES OF THE UNITED STATES OF AMERICA*
  Jun, G., Manning, A., Almeida, M., Zawistowski, M., Wood, A. R., Teslovich, T. M., Fuchsberger, C., Feng, S., Cingolani, P., Gaulton, K. J., Dyer, T., Blackwell, T. W., Chen, et al
  2018; 115 (2): 379–84

- **Protein-altering variants associated with body mass index implicate pathways that control energy intake and expenditure in obesity** *NATURE GENETICS*
  Turcot, V., Lu, Y., Highland, H. M., Schurmann, C., Justice, A. E., Fine, R. S., Bradfield, J. P., Esko, T., Giri, A., Graff, M., Guo, X., Hendricks, A. E., Karaderi, et al
  2018; 50 (1): 26-+

- **Protein-altering variants associated with body mass index implicate pathways that control energy intake and expenditure in obesity.** *Nature genetics*
  Turcot, V. n., Lu, Y. n., Highland, H. M., Schurmann, C. n., Justice, A. E., Fine, R. S., Bradfield, J. P., Esko, T. n., Giri, A. n., Graff, M. n., Guo, X. n., Hendricks, A. E., Karaderi, et al
  2018; 50 (1): 26–41

- **Data Descriptor: Sequence data and association statistics from 12,940 type 2 diabetes cases and controls** *SCIENTIFIC DATA*
  Flannick, J., Fuchsberger, C., Mahajan, A., Teslovich, T. M., Agarwala, V., Gaulton, K. J., Caulkins, L., Koesterer, R., Ma, C., Moutsianas, L., McCarthy, D. J., Rivas, M. A., Perry, et al
  2017; 4: 170179

- **Large-scale GWAS identifies multiple loci for hand grip strength providing biological insights into muscular fitness** *NATURE COMMUNICATIONS*
  Willems, S. M., Wright, D. J., Day, F. R., Trajanoska, K., Joshi, P. K., Morris, J. A., Matteini, A. M., Garton, F. C., Grarup, N., Oskolkov, N., Thalamuthu, A., Mangino, M., Liu, et al
  2017; 8: 16015

- **Mosaic mutations in blood DNA sequence are associated with solid tumor cancers** *NPJ GENOMIC MEDICINE*
  Artomov, M., Rivas, M. A., Genovese, G., Daly, M. J.
  2017; 2: 22

- **biMM: Efficient estimation of genetic variances and covariances for cohorts with high-dimensional phenotype measurements.** *Bioinformatics*
  Pirinen, M., Benner, C., Marttinen, P., Järvelin, M., Rivas, M. A., Ripatti, S.
  2017

- **Variant Enriched in the Finnish Population is Associated With Fasting Insulin Levels and Type 2 Diabetes Risk.** *Diabetes*
  Manning, A., Highland, H. M., Gasser, J., Sim, X., Tukiainen, T., Fontanillas, P., Grarup, N., Rivas, M. A., Mahajan, A., Locke, A. E., Cingolani, P., Pers, T. H., Viñuela, et al
  2017

- **Rare and low-frequency coding variants alter human adult height.** *Nature*
  Marouli, E., Graff, M., Medina-Gomez, C., Lo, K. S., Wood, A. R., Kjaer, T. R., Fine, R. S., Lu, Y., Schurmann, C., Highland, H. M., Rüeger, S., Thorleifsson, G., Justice, et al
  2017; 542 (7640): 186-190

- **Frameshift indels introduced by genome editing can lead to in-frame exon skipping.** *PloS one*
  Lalonde, S., Stone, O. A., Lessard, S., Lavertu, A., Desjardins, J., Beaudoin, M., Rivas, M., Stainier, D. Y., Lettre, G.
  2017; 12 (6)

- **Landscape of X chromosome inactivation across human tissues.** *Nature*
  Tukiainen, T. n., Villani, A. C., Yen, A. n., Rivas, M. A., Marshall, J. L., Satija, R. n., Aguirre, M. n., Gauthier, L. n., Fleharty, M. n., Kirby, A. n., Cummings, B. B., Castel, S. E., Karczewski, et al
  2017; 550 (7675): 244–48

- **Analysis of protein-coding genetic variation in 60,706 humans** *NATURE*
  Lek, M., Karczewski, K. J., Minikel, E. V., Samocha, K. E., Banks, E., Fennell, T., O'Donnell-Luria, A. H., Ware, J. S., Hill, A. J., Cummings, B. B., Tukiainen, T., Birnbaum, D. P., Kosmicki, et al
  2016; 536 (7616): 285-?

- **The genetic architecture of type 2 diabetes** *NATURE*
  Fuchsberger, C., Flannick, J., Teslovich, T. M., Mahajan, A., Agarwala, V., Gaulton, K. J., Ma, C., Fontanillas, P., Moutsianas, L., McCarthy, D. J., Rivas, M. A., Perry, J. R., Sim, et al
  2016; 536 (7614): 41-?

- **A null mutation in ANGPTL8 does not associate with either plasma glucose or type 2 diabetes in humans** *BMC ENDOCRINE DISORDERS*
  Clapham, K. R., Chu, A. Y., Wessel, J., Natarajan, P., Flannick, J., Rivas, M. A., Sartori, S., Mehran, R., Baber, U., Fuster, V., Scott, R. A., Rader, D. J., Boehnke, et al
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