BIOGRAPHICAL SKETCH

NAME: RIVAS, MANUEL

eRA COMMONS USER NAME (credential, e.g., agency login): rivas.manuel

POSITION TITLE: Assistant Professor

EDUCATION/TRAINING *(Begin with baccalaureate or other initial professional education, such as nursing, include postdoctoral training and residency training if applicable.)*

INSTITUTION AND LOCATION	DEGREE (if applicable)	START DATE MM/YYYY	END DATE MM/YYYY	FIELD OF STUDY
Massachusetts Institute of Technology, Cambridge, MA	BS	09/2004	06/2008	Mathematics
University of Oxford, Oxford	DPHIL	09/2010	11/2015	Clinical Medicine

Contact information

Department of Biomedical Data Science
Stanford School of Medicine
365 Lasuen Street, Littlefield Room 337
Stanford, CA 94305

Contact Number: +1-650-724-6077
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Personal Statement

I am a biomedical data scientist whose research focuses on developing statistical methods and computational tools to analyze massive human genetic datasets to address fundamental questions in medicine and biology. Our current research concentrates on four themes: 1) generating effective therapeutic hypotheses from human genetic data; 2) developing technologies for integrated learning healthcare systems; 3) inferring the global distribution of common and rare disease predisposition genes; and 4) developing statistical learning models and optimization algorithms. Examples of our research accomplishments include:

- Identification of protective protein truncating genetic variants against crohn's disease and ulcerative colitis (CARD9 and RNF186), which are being pursued as therapeutic hypotheses;
- development of statistical methods for rare variant association studies across large-scale genome sequencing study designs;
- development of statistical methods for the analysis of RNA-sequencing data;
- and development of error models and tools for processing of high-throughput sequencing data.

My lab has dry lab expertise and students and post-docs from diverse fields including: machine learning, natural language processing, biomedical informatics, statistics, and human genetics. I serve as an advisor to startups in the areas of applied statistics, biomedical data science, and human genetics. I also serve as a member of a diversity and inclusion in scientific computing committee in the non-profit organization NumFOCUS.

Selected Publications

1. Neale BM, Rivas MA, Voight BF, Altshuler D, Devlin B, Orho-Melander M, Kathiresan S, Purcell SM, Roeder K, Daly MJ. Testing for an unusual distribution of rare variants. PLoS Genet. 2011 Mar;7(3):e1001322. PubMed PMID: 21408211; PubMed Central PMCID: PMC3048375.
2. Rivas MA, Graham D, Sulem P, Stevens C, Desch AN, Goyette P, Gudbjartsson D, Jonsdottir I, Thorsteinsdottir U, Degenhardt F, Mucha S, Kurki MI, Li D, D'Amato M, Annese V, Vermeire S, Weersma RK, Halfvarson J, Paavola-Sakki P, Lappalainen M, Lek M, Cummings B, Tukiainen T, Haritunians T, Halme L, Koskinen LL, Ananthakrishnan AN, Luo Y, Heap GA, Visschedijk MC, MacArthur DG, Neale BM, Ahmad T, Anderson CA, Brant SR, Duerr RH, Silverberg MS, Cho JH,

Palotie A, Saavalainen P, Kontula K, Färkkilä M, McGovern DP, Franke A, Stefansson K, Rioux JD, Xavier RJ, Daly MJ, Barrett J, de Lane K, Edwards C, Hart A, Hawkey C, Jostins L, Kennedy N, Lamb C, Lee J, Lees C, Mansfield J, Mathew C, Mowatt C, Newman B, Nimmo E, Parkes M, Pollard M, Prescott N, Randall J, Rice D, Satsangi J, Simmons A, Tremelling M, Uhlig H, Wilson D, Abraham C, Achkar JP, Bitton A, Boucher G, Croitoru K, Fleshner P, Glas J, Kugathasan S, Limbergen JV, Milgrom R, Proctor D, Regueiro M, Schumm PL, Sharma Y, Stempak JM, Targan SR, Wang MH. A protein-truncating R179X variant in RNF186 confers protection against ulcerative colitis. Nat Commun. 2016 Aug 9;7:12342. PubMed PMID: 27503255; PubMed Central PMCID: PMC4980482.

3. Rivas MA, Pirinen M, Conrad DF, Lek M, Tsang EK, Karczewski KJ, Maller JB, Kukurba KR, DeLuca DS, Fromer M, Ferreira PG, Smith KS, Zhang R, Zhao F, Banks E, Poplin R, Ruderfer DM, Purcell SM, Tukiainen T, Minikel EV, Stenson PD, Cooper DN, Huang KH, Sullivan TJ, Nedzel J, Bustamante CD, Li JB, Daly MJ, Guigo R, Donnelly P, Ardlie K, Sammeth M, Dermitzakis ET, McCarthy MI, Montgomery SB, Lappalainen T, MacArthur DG. Human genomics. Effect of predicted protein-truncating genetic variants on the human transcriptome. Science. 2015 May 8;348(6235):666-9. PubMed PMID: 25954003; PubMed Central PMCID: PMC4537935.
4. Rivas MA, Beaudoin M, Gardet A, Stevens C, Sharma Y, Zhang CK, Boucher G, Ripke S, Ellinghaus D, Burtt N, Fennell T, Kirby A, Latiano A, Goyette P, Green T, Halfvarson J, Haritunians T, Korn JM, Kuruvilla F, Lagacé C, Neale B, Lo KS, Schumm P, Törkvist L, Dubinsky MC, Brant SR, Silverberg MS, Duerr RH, Altshuler D, Gabriel S, Lettre G, Franke A, D'Amato M, McGovern DP, Cho JH, Rioux JD, Xavier RJ, Daly MJ. Deep resequencing of GWAS loci identifies independent rare variants associated with inflammatory bowel disease. Nat Genet. 2011 Oct 9;43(11):1066-73. PubMed PMID: 21983784; PubMed Central PMCID: PMC3378381.

B. Positions and Honors

Positions and Employment

2008 - 2010	Bioinformatics Analyst, Broad Institute of MIT and Harvard, Medical and Population Genetics, Cambridge, MA
2014 - 2016	Computational Scientist, Broad Institute of MIT and Harvard, Medical and Population Genetics, Cambridge, MA
2016 -	Assistant Professor, Stanford University, Department of Biomedical Data Science, School of Medicine, Stanford, CA

Other Experience and Professional Memberships

2016 -	Member, Stanford Bio-X
2016 -	Member, International Society for Bayesian Analysis
2016 -	Member, American Society of Human Genetics
2017 -	Member Diversity and Inclusion in Scientific Computing Committee, NUMFOCUS

Consulting and Industry Service

2016 -	Consultant in Statistical Genomics, Genomics PLC (Oxford, UK)
2017 -	Consultant in Applied Statistics, Prime Genomics (Santa Cruz, California, US)

Honors

2004 - 2008	Gates Millennium scholarship, Bill and Melinda Gates Foundation
2004	Exxon-Mobil State and National Scholar, Exxon-Mobil
2004	Espiritu de Superacion scholastic award, Ford Foundation
2004	Advanced Placement National Scholar, College Board
2010	NMD Osler Award Winner, University of Oxford
2010	Clarendon Scholarship, University of Oxford
2016	NMD Graduate Prize Winner, University of Oxford

Publications (47 total)

Journal Article (46)

2017

1. Manning A, Highland HM, Gasser J, Sim X, Tukiainen T, Fontanillas P, Grarup N, **Rivas MA**, …, Gloyn AL, Lindgren CM. A low-frequency inactivating *AKT2* variant enriched in the Finnish population Is associated with fasting insulin levels and type 2 diabetes risk. **Diabetes**. 2017; 66(7):2019-2032. NIHMSID: EMS72347 PubMed PMID: 28341696, PMCID: PMC5482074.

2. Pirinen M, Benner C, Marttinen P, Järvelin MR, **Rivas MA**, Ripatti S. biMM: Efficient estimation of genetic variances and covariances for cohorts with high-dimensional phenotype measurements. **Bioinformatics**. 2017; PubMed PMID: 28369165.

3. Marouli E, Graff M, Medina-Gomez C, Lo KS, Wood AR, Kjaer TR, Fine RS, Lu Y, Schurmann C, Highland HM, Rüeger S, Thorleifsson G, Justice AE, Lamparter D, Stirrups KE, Turcot V, Young KL, Winkler TW, Esko T, Karaderi T, Locke AE, Masca NG, Ng MC, Mudgal P, **Rivas MA**, …, Hirschhorn JN, Deloukas P, Lettre G. Rare and low-frequency coding variants alter human adult height. Nature. 2017; 542(7640):186-190. NIHMSID: NIHMS834200 PubMed PMID: 28146470, PMCID: PMC5302847.

4. Lalonde S, Stone OA, Lessard S, Lavertu A, Desjardins J, Beaudoin M, **Rivas M**, Stainier DYR, Lettre G. Frameshift indels introduced by genome editing can lead to in-frame exon skipping. **PloS one**. 2017; 12(6):e0178700. PubMed PMID: 28570605, PMCID: PMC5453576.

2016

5. Graham DB, Lefkovith A, …, **Rivas MA**, Daly MJ, Carr SA, Wijmenga C, Xavier RJ. TMEM258 Is a component of the Oligosaccharyltransferase Complex Controlling ER Stress and Intestinal Inflammation. **Cell reports**. 2016;17(11):2955-2965. PubMed PMID: 27974209.

6. Chuang LS, Villaverde N, Hui KY, Mortha A, Rahman A, Levine AP, Haritunians T, Evelyn Ng SM, Zhang W, Hsu NY, Facey JA, Luong T, Fernandez-Hernandez H, Li D, **Rivas M**, …, Peter I, Cho JH. A frameshift in CSF2RB predominant among Ashkenazi Jews increases risk for Crohn's disease and reduces monocyte signaling via GM-CSF. **Gastroenterology**. 2016; 151(4):710-723.e2. NIHMSID: NIHMS800998 PubMed PMID: 27377463,PMCID: PMC5037012.

7. Ferreira PG, Oti M, Barann M, Wieland T, Ezquina S, Friedländer MR, **Rivas MA**, Esteve-Codina A, Rosenstiel P, Strom TM, Lappalainen T, Guigó R, Sammeth M. Sequence variation between 462 human individuals fine-tunes functional sites of RNA processing. **Scientific reports**. 2016; 6:32406. PubMed PMID: 27617755, PMCID: PMC5019111.

8. Lek M, Karczewski KJ, Minikel EV, Samocha KE, Banks E, Fennell T, O'Donnell-Luria AH, Ware JS, Hill AJ, Cummings BB, Tukiainen T, Birnbaum DP, Kosmicki JA, Duncan LE, Estrada K, Zhao F, Zou J, Pierce-Hoffman E, Berghout J, Cooper DN, Deflaux N, DePristo M, Do R, Flannick J, Fromer M, Gauthier L, Goldstein J, Gupta N, Howrigan D, Kiezun A, Kurki MI, Moonshine AL, Natarajan P, Orozco L, Peloso GM, Poplin R, **Rivas MA**, …, Daly MJ, MacArthur DG. Analysis of protein-coding genetic variation in 60,706 humans. **Nature**. 2016; 536(7616):285-91. NIHMSID: NIHMS798561 PubMed PMID: 27535533, PMCID: PMC5018207.

9. **Rivas MA**, Graham D, Sulem P, Stevens C, Desch AN, Goyette P, Gudbjartsson D,Jonsdottir I, Thorsteinsdottir U, …, Xavier RJ, Daly MJ. A protein-truncating R179X variant in RNF186 confers protection against ulcerative colitis. **Nature communications**. 2016; 7:12342. PubMed PMID: 27503255, PMCID: PMC4980482.

10. Fuchsberger C, Flannick J, Teslovich TM, Mahajan A, Agarwala V, Gaulton KJ, Ma C, Fontanillas P, Moutsianas L, McCarthy DJ, **Rivas MA**, …, Boehnke M, Altshuler D, McCarthy MI. The genetic architecture of type 2 diabetes. **Nature**. 2016; 536(7614):41-7. NIHMSID: EMS68809 PubMed PMID: 27398621, PMCID: PMC5034897.

11. Kosmicki JA, Churchhouse CL, **Rivas MA**, Neale BM. Discovery of rare variants for complex phenotypes. **Human genetics**. 2016; 135(6):625-34. PubMed PMID: 27221085.

12. Clapham KR, Chu AY, Wessel J, Natarajan P, Flannick J, **Rivas MA**, …, Kathiresan S, Peloso GM. A null mutation in ANGPTL8 does not associate with either plasma glucose or type 2 diabetes in humans. **BMC endocrine disorders**. 2016; 16:7. PubMed PMID: 26822414, PMCID: PMC4730725.

13. Visschedijk MC, Alberts R, Mucha S, Deelen P, …, **Rivas MA**, Franke A, van Diemen CC, Weersma RK. Pooled Resequencing of 122 Ulcerative Colitis Genes in a Large Dutch Cohort Suggests Population-Specific Associations of Rare Variants in MUC2. **PloS one**. 2016; 11(8):e0159609. PubMed [journal] PMID: 27490946, PMCID: PMC4973970.

14. Richardson TG, Shihab HA, **Rivas MA**, McCarthy MI, Campbell C, Timpson NJ, Gaunt TR. A Protein Domain and Family Based Approach to Rare Variant Association Analysis. **PloS one**. 2016; 11(4):e0153803. PubMed [journal] PMID: 27128313, PMCID: PMC4851355.

2015

15. Kim YJ, Lee J, Kim BJ, Park T. A new strategy for enhancing imputation quality of rare variants from next-generation sequencing data via combining SNP and exome chip data. **BMC genomics**. 2015; 16:1109. PubMed PMID: 26715385, PMCID: PMC4696174

16. **GTEx consortium.** A Novel Approach to High-Quality Postmortem Tissue Procurement: The GTEx Project. **Biopreservation and biobanking**. 2015; 13(5):311-9. PubMed PMID: 26484571, PMCID: PMC467518.

17. Pirinen M, Lappalainen T, Zaitlen NA, Dermitzakis ET, Donnelly P, McCarthy MI, **Rivas MA**. Assessing allele-specific expression across multiple tissues from RNA-seq read data. **Bioinformatics**. 2015; 31(15):2497-504. PubMed PMID: 25819081, PMCID: PMC4514921

18. Baran Y, Subramaniam M, Biton A, Tukiainen T, Tsang EK, **Rivas MA**, …, Zaitlen NA, Lappalainen T. The landscape of genomic imprinting across diverse adult human tissues. **Genome research**. 2015; 25(7):927-36. PubMed PMID: 25953952, PMCID: PMC4484390.

19. **Rivas MA**, Pirinen M, Conrad DF, Lek M, Tsang EK, Karczewski KJ, Maller JB, …, Lappalainen T, MacArthur DG. Human genomics. Effect of predicted protein-truncating genetic variants on the human transcriptome. **Science**. 2015; 348(6235):666-9. NIHMSID: NIHMS712687 PubMed PMID: 25954003, PMCID: PMC4537935.

20. **GTEx consortium.** The Genotype-Tissue Expression (GTEx) pilot analysis: multi-tissue gene regulation in humans. **Science**. 2015; 348(6235):648-60. NIHMSID: NIHMS712356 PubMed [journal] PMID: 25954001, PMCID: PMC4547484.

21. Pierson E, Koller D, Battle A, Mostafavi S, Ardlie KG, Getz G, Wright FA, **GTEx consortium**, Kellis M, Volpi S, Dermitzakis ET. Sharing and specificity of co-expression networks across 35 human tissues. **PLoS computational biology**. 2015; 11(5):e1004220. PubMed PMID: 25970446, PMCID: PMC4430528.

22. Moutsianas L, Agarwala V, Fuchsberger C, Flannick J, **Rivas MA**, Gaulton KJ, Albers PK, McVean G, Boehnke M, Altshuler D, McCarthy MI. The power of gene-based rare variant methods to detect disease-associated variation and test hypotheses about complex disease. **PLoS genetics**. 2015; 11(4):e1005165. PubMed PMID:25906071, PMCID: PMC4407972

23. Wood AR, Tuke MA, Nalls M, Hernandez D, Gibbs JR, Lin H, Xu CS, Li Q, Shen J, Jun G, Almeida M, Tanaka T, Perry JR, Gaulton K, **Rivas M**, …, Frayling TM. Whole-genome sequencing to understand the genetic architecture of common gene expression and biomarker phenotypes. **Human molecular genetics**. 2015; 24(5):1504-12. PubMed PMID: 25378555, PMCID: PMC4321449.

24. Do R, Stitziel NO, Won HH, Jørgensen AB, Duga S, Angelica Merlini P, Kiezun A, Farrall M, Goel A, Zuk O, Guella I, Asselta R, …, **Rivas MA**, Donnelly P, …, Altshuler D, Gabriel S, Kathiresan S. Exome sequencing identifies rare LDLR and APOA5 alleles conferring risk for myocardial infarction. **Nature**. 2015; 518(7537):102-6. NIHMSID: NIHMS633391 PubMed PMID: 25487149, PMCID: PMC4319990

25. Mahajan A, Sim X, Ng HJ, Manning A, **Rivas MA**, Highland HM, …, Altshuler D, Meigs JB, Boehnke M, McCarthy MI, Lindgren CM, Gloyn AL. Identification and functional characterization of G6PC2 coding variants influencing glycemic traits define an effector transcript at the G6PC2-ABCB11 locus. **PLoS genetics**. 2015;11(1):e1004876. PubMed PMID: 25625282, PMCID: PMC4307976.

2014

26. Majithia AR, Flannick J, Shahinian P, Guo M, Bray MA, **GoT2D consortium**, Fontanillas P, Gabriel SB, Rosen ED, Altshuler D. Rare variants in PPARG with decreased activity in adipocyte differentiation are associated with increased risk of type 2 diabetes. **Proceedings of the National Academy of Sciences of the United States of America**. 2014; 111(36):13127-32. PubMed PMID: 25157153, PMCID: PMC4246964.

27. Lange LA, Hu Y, Zhang H, Xue C, Schmidt EM, Tang ZZ, Bizon C, Lange EM, Smith JD, Turner EH, Jun G, Kang HM, Peloso G, Auer P, Li KP, Flannick J, Zhang J, Fuchsberger C, Gaulton K, Lindgren C, Locke A, Manning A, Sim X, **Rivas MA**, …, Willer CJ. Whole-exome sequencing identifies rare and low-frequency coding variants associated with LDL cholesterol. **American journal of human genetics**. 2014; 94(2):233-45. PubMed PMID: 24507775, PMCID: PMC3928660.

28. McCarthy DJ, Humburg P, Kanapin A, **Rivas MA**, Gaulton K, Cazier JB, Donnelly P. Choice of transcripts and software has a large effect on variant annotation. **Genome medicine**. 2014; 6(3):26. PubMed PMID: 24944579, PMCID: PMC4062061.

2013

29. **Rivas MA**, Pirinen M, Neville MJ, Gaulton KJ, Moutsianas L, Lindgren CM, Karpe F, McCarthy MI, Donnelly P. Assessing association between protein truncating variants and quantitative traits. **Bioinformatics**. 2013; 29(19):2419-26. PubMed PMID: 23860716, PMCID: PMC3777107.

30. Lappalainen T, Sammeth M, Friedländer MR, 't Hoen PA, Monlong J, **Rivas MA**, Gonzàlez-Porta M, …, Dermitzakis ET. Transcriptome and genome sequencing uncovers functional variation in humans. **Nature**. 2013; 501(7468):506-11. NIHMSID: NIHMS512974 PubMed PMID: 24037378, PMCID: PMC3918453.

31. Ellinghaus D, Zhang H, Zeissig S, Lipinski S, Till A, Jiang T, Stade B, Bromberg Y, Ellinghaus E, Keller A, **Rivas MA**, …, Parkes M, Franke A. Association between variants of PRDM1 and NDP52 and Crohn's disease, based on exome sequencing and functional studies. **Gastroenterology.** 2013; 145(2):339-47. NIHMSID: NIHMS500460 PubMed PMID: 23624108, PMCID: PMC3753067.

32. Ruark E, Snape K, Humburg P, Loveday C, Bajrami I, Brough R, Rodrigues DN, Renwick A, Seal S, Ramsay E, Duarte Sdel V, **Rivas MA**, …, Donnelly P, Rahman N. Mosaic *PPM1D* mutations are associated with predisposition to breast and ovarian cancer. **Nature**. 2013; 493(7432):406-10. NIHMSID: EMS54398 PubMed PMID: 23242139, PMCID: PMC3759028.

33. Fu W, O'Connor TD, Jun G, Kang HM, **ESP consortium**, Abecasis G, Leal SM, Gabriel S, Rieder MJ, Altshuler D, Shendure J, Nickerson DA, Bamshad MJ, Akey JM. Analysis of 6,515 exomes reveals the recent origin of most human protein-coding variants. **Nature**. 2013; 493(7431):216-20. NIHMSID: NIHMS416429 PubMed PMID: 23201682, PMCID: PMC3676746.

34. Diogo D, Kurreeman F, Stahl EA, Liao KP, Gupta N, Greenberg JD, **Rivas MA**, …, Raychaudhuri S, Plenge RM. Rare, low-frequency, and common variants in the protein-coding sequence of biological candidate genes from GWASs contribute to risk of rheumatoid arthritis. **American journal of human genetics**. 2013; 92(1):15-27. PubMed PMID: 23261300, PMCID: PMC3542467.

35. Clarke GM, **Rivas MA**, Morris AP. A flexible approach for the analysis of rare variants allowing for a mixture of effects on binary or quantitative traits. **PLoS genetics**. 2013; 9(8):e1003694. PubMed PMID: 23966874, PMCID: PMC3744430.

36. Beaudoin M, Goyette P, Boucher G, Lo KS, **Rivas MA**, …, Daly MJ, Rioux JD. Deep resequencing of GWAS loci identifies rare variants in *CARD9*, *IL23R* and *RNF186* that are associated with ulcerative colitis. **PLoS genetics**. 2013; 9(9):e1003723. PubMed PMID: 24068945, PMCID: PMC3772057.

2012

37. Beaudoin M, Lo KS, N'Diaye A, **Rivas MA**, Dubé MP, Laplante N, Phillips MS, Rioux JD, Tardif JC, Lettre G. Pooled DNA resequencing of 68 myocardial infarction candidate genes in French canadians. **Circulation**. 2012; 5(5):547-54. PubMed PMID: 22923420.

38. Ameur A, Enroth S, Johansson A, Zaboli G, Igl W, Johansson AC, **Rivas MA**, Daly MJ, Schmitz G, Hicks AA, Meitinger T, Feuk L, van Duijn C, Oostra B, Pramstaller PP, Rudan I, Wright AF, Wilson JF, Campbell H, Gyllensten U. Genetic adaptation of fatty-acid metabolism: a human-specific haplotype increasing the biosynthesis of long-chain omega-3 and omega-6 fatty acids. **American journal of human genetics**. 2012; 90(5):809-20. PubMed PMID: 22503634, PMCID: PMC3376635.

2011

39. **Rivas MA**, Beaudoin M, Gardet A, Stevens C, Sharma Y, Zhang CK, Boucher G, Ripke S, …, Xavier RJ, Daly MJ. Deep resequencing of GWAS loci identifies independent rare variants associated with inflammatory bowel disease. **Nature genetics**. 2011; 43(11):1066-73. NIHMSID: NIHMS335188 PubMed PMID: 21983784, PMCID: PMC3378381.

40. Danecek P, Auton A, Abecasis G, Albers CA, Banks E, DePristo MA, Handsaker RE, Lunter G, **1000 Genomes Data Processing subgroup**, Marth GT, Sherry ST, McVean G, Durbin R. The variant call format and VCFtools. **Bioinformatics**. 2011; 27(15):2156-8. PubMed PMID: 21653522, PMCID: PMC3137218.

41. DePristo MA, Banks E, Poplin R, Garimella KV, Maguire JR, Hartl C, Philippakis AA, del Angel G, **Rivas MA**, …, Altshuler D, Daly MJ. A framework for variation discovery and genotyping using next-generation DNA sequencing data. **Nature genetics**. 2011; 43(5):491-8. NIHMSID: NIHMS281651 PubMed PMID: 21478889, PMCID: PMC3083463.

42. Neale BM, **Rivas MA**, Voight BF, Altshuler D, Devlin B, Orho-Melander M, Kathiresan S, Purcell SM, Roeder K, Daly MJ. Testing for an unusual distribution of rare variants. **PLoS genetics**. 2011; 7(3):e1001322. PubMed [journal] PMID: 21408211, PMCID: PMC3048375.

2010

43. **1000 Genomes Project consortium**. A map of human genome variation from population-scale sequencing. **Nature**. 2010; 467(7319):1061-73. NIHMSID: UKMS34220 PubMed PMID: 20981092, PMCID: PMC3042601.

44. Calvo SE, Tucker EJ, Compton AG, Kirby DM, Crawford G, Burtt NP, **Rivas M**, Guiducci C, Bruno DL, Goldberger OA, …, Gabriel SB, Daly MJ, Thorburn DR, Mootha VK. High-throughput, pooled sequencing identifies mutations in NUBPL and FOXRED1 in human complex I deficiency. **Nature genetics**. 2010; 42(10):851-8. NIHMSID: NIHMS228290 PubMed PMID: 20818383, PMCID: PMC2977978.

45. Kirby A, Kang HM, Wade CM, Cotsapas C, Kostem E, Han B, Furlotte N, Kang EY, **Rivas M**, Bogue MA, Frazer KA, Johnson FM, Beilharz EJ, Cox DR, Eskin E, Daly MJ. Fine mapping in 94 inbred mouse strains using a high-density haplotype resource. **Genetics**. 2010; 185(3):1081-95. PubMed PMID: 20439770, PMCID: PMC2907194.

2008

46. Choy E, Yelensky R, Bonakdar S, Plenge RM, Saxena R, De Jager PL, Shaw SY, Wolfish CS, Slavik JM, Cotsapas C, **Rivas M**, Dermitzakis ET, Cahir-McFarland E, Kieff E, Hafler D, Daly MJ, Altshuler D. Genetic analysis of human traits in vitro: drug response and gene expression in lymphoblastoid cell lines. **PLoS genetics**. 2008; 4(11):e1000287. PubMed PMID: 19043577, PMCID: PMC2583954.

Book Section (1)

1. **Rivas MA** and Moutsianas L. Power of rare variant aggregate tests in *Assessing rare variation in complex traits*. 185-199 (2015). Springer, New York.

Additional Information: Research Support and/or Scholastic Performance

Other Experience and Professional Memberships

2017 -	Member, Diversity and Inclusion in Scientific Computing Committee (DISC), NUMFOCUS
2016 -	Member, NHGRI's Genome Sequencing Program Analysis Committee
2016 -	Member, NHGRI's Genome Sequencing Program Steering Committee
2016 -	Member, NHGRI's Genome Sequencing Program Data Coordinating Center
2016 -	Member, Stanford Bio-X
2016 -	Member, International Society for Bayesian Analysis
2016 -	Member, American Society of Human Genetics
2013-2015	Member, Genotype Tissue Expression Project Analysis Commitee

Research Support

Ongoing Research Support

Educational and Outreach Activities

Post-doctoral fellows trained
1. Dr. Christopher Mark DeBoever (9/2016-current)
2. Dr. Johanne Marie Justesen (11/2017-current)

Graduate students trained
1. Yosuke Tanigawa (9/2016-current, joint with Prof. Gil Bejerano)
2. Oliver Bear Don't Walk IV (9/2016-6/2017, Master's student), now PhD student at Columbia University
3. Ananth Ganesan (4/2017-6/2017)
4. Adam Lavertu (9/2016-12/2016), now PhD student with Russ Altman
5. Mamie Wang (9/2016-12/2016).
6. Greg McInnes (1/2017-4/2017), now PhD student with Russ Altman

Staff scientists trained
1. Mr. Matthew Aguirre (9/2017-current)

Visiting scientists trained
1. Dr. Anna Cichonska (8/2017-9/2017)

Courses Taught
BIODS215: Topics in Biomedical Data Science: Large-scale inference (S2017)
GENE245: Statistical and Machine Learning Methods for Genomics (S2017, Guest lecture)
BIODS260C: Workshop in Biostatistics

Invited Conferences, Keynote Lectures, and Departmental Seminars (44 total)

2017

44. General session at INMEGEN, Mexico City, Mexico (August 2017)
43. Oxford Big Data Institute, Oxford, UK (July 2017)
42. Novo Nordisk Foundation Meeting, Stanford, California, USA (June 2017)
41. 23andMe Genome Research Day, Mountain View, California, USA (June 2017)
40. Think Different Seminar Series, Helsinki, Finland (March 2017)
39. BMIR research colloquium, Stanford, California, USA (February 2017)
38. Genetics research consortium meeting, Biogen, Cambridge, Massachusetts, USA (January 2017)

2016

37. UNAN-Managua, Managua, Nicaragua (December 2016)
36. Target Validation using Genomics and Informatics, EMBL-EBI, Heidelberg, Germany (December 2016)
35. Institute for Computational and Mathematical Engineering First Year Seminar Series, Stanford, California, USA (November 2016)
34. Genomics of Common Diseases, Bethesda, Maryland, USA (September 2016)
33. Medical and Population Genetics, Broad Institute, Cambridge, Massachusetts, USA (April 2016)
32. Vertex seminar, Boston, Massachusetts, USA (April 2016)
31. Harvard School of Public Health Program in Quantitative Genomics Seminar, Cambridge, Massachusetts, USA (April 2016)
30. Stanford University seminar, Stanford, California, USA (April 2016)
29. Genomics PLC seminar, Oxford, UK (April 2016)
28. Wellcome Trust Centre for Human Genetics seminar, Oxford, UK (April 2016)

2015

27. MIT Online Science, Technology, and Engineering Community (MOSTEC) program**.** Cambridge, Massachusetts, USA (December, 2015)
26. Digestive Disease Week, Washington, DC, USA (May, 2015)
25. Harvard School of Public Health Program in Quantitative Genomics Working Group Series. Boston, Massachusetts, USA (January, 2015)
24. Nordic Information for Action eScience Center, Helsinki, Finland (January, 2015)
23. Nordic Information for Action eScience Center, GWAS Mini Course, Helsinki, Finland (January, 2015)

2014

22. PyData NYC, New York, USA (November, 2014)
21. Boston eQTL meeting, Cambridge, USA (June, 2014)
20. 1000 Genomes Project and Beyond, Cambridge, UK (June, 2014)
19. GTEx Community Meeting, Cambridge, Massachusetts, USA (June, 2014)
18. Biology of Genomes, Cold Spring Harbor, New York, USA (May, 2014)
17. The Jackson Laboratory Genomic Medicine Seminar, Farmington, Connecticut, USA (January, 2014)

2013

16. Cardio + Medicine Seminar, Stanford, California, USA (November, 2013)
15. UCLA Bioinformatics Seminar, Los Angeles, California, USA (November, 2013)
14. Genomic Medicine in the Mediterranean: GM2**,** Crete, Greece (October, 2013)
13. Leena Peltonen School of Human Genomics, Sanger Institute, Cambridge, UK (August, 2013)
12. Stanford University Seminar, Stanford University, Stanford, California, USA (June, 2013)
11. GTEx Community Workshop Meeting, Cambridge, Massachusetts, USA (June, 2013)
10. European Society of Human Genetics, Paris, France (June, 2013)

9. University of Liege seminar, Liege, Belgium (April, 2013)
8. Institute of Cancer Research seminar, London, England (February, 2013)

2011

7. Genomics of Common Diseases, Cambridge, UK (September, 2011)
6. International Congress of Human Genetics, Montreal, Canada (November, 2011)

2010

5. MathGenBio Seminar, Department of Statistics, University of Oxford, Oxford, UK (November 2010)
4. American Society of Human Genetics Conference**,** Washington, D.C. (November 2010)
3. Department of Computer Science seminar**,** Columbia University, New York, NY, USA (July, 2010)
2. Sanger Institute seminar**,** Cambridge, UK (January, 2010)
1. Stochastic Seminar, Massachusetts Institute of Technology, Cambridge, MA, USA (January, 2010)