Ryan E Mills, Ph.D.
Assistant Professor, Department of Computational Medicine & Bioinformatics
Assistant Professor, Department of Human Genetics
University of Michigan Medical School
100 Washtenaw Ave, Room 2055B
Phone: 734-647-9628
Email: remills@med.umich.edu

Education and Training

Education

08/1996-05/2000	AB, Biology, Wabash College, Crawfordsville, IN
08/2000-08/2003	MS, Applied Biology, Georgia Institute of Technology, Atlanta, GA
09/2003-05/2006	PhD, Bioinformatics, Georgia Institute of Technology, Atlanta, GA

Academic, Administrative, Clinical and Military Appointments

Academic Appointments

06/2006-10/2008	Postdoctoral Fellow, Emory University, Atlanta, GA
11/2008-12/2011	Research Associate, Brigham & Women's Hospital, Harvard Medical School, Boston, MA
01/2012-present	Assistant Professor, Human Genetics, University of Michigan - Ann Arbor, Ann Arbor, MI
01/2012-present	Assistant Professor, Computational Medicine & Bioinformatics, University of Michigan - Ann Arbor, Ann Arbor, MI

Administrative Appointments

01/2009-12/2011	Team Leader, Bioinformatics and Medical Diagnostics Team, Molecular Genetic Research Unit, Brigham and Women's Hospital, Boston, MA

Research Interests

- The primary purpose of sequencing genomes is to identify the underlying genetic variation between individuals and to explore what role those changes have on human phenotypes. Our research laboratory develops and implements methods to precisely identify and resolve different types of genomic variation. Our goal is to integrate this information with other forms of biologically and medically relevant data to improve our overall understanding of human health and disease.

Grants

Current Grants

5 R01 AI118886-05: Fidelity, robustness, and diversity in RNA virus evolution and pathogenesis NIH-DHHS-US-15-PAF06122
Co-I with Effort (Principal Investigator: Lauring, Adam)
01/2016-12/2020. $2,023,562 ($397,708)

5 R01 HG007068-04: Discovery and analysis of structural variation in whole genome sequences NIH-DHHS-US-13-PAF00068
Mills, Ryan Edward, PI
09/2013-07/2018. $1,526,575 ($382,699)

1 U41 HG007497-01: An Integrative Analysis of Structural Variation for the 1000 Genomes Project
SubK-NIH-DHHS-US through a consortium with Jackson Laboratory- 13-PAF01456
Mills, Ryan Edward, PI
09/2013-08/2018. $397,471 ($94,527)

5 U01 MH106892-05: 2/3 Schizophrenia Genetics and Brain Somatic Mosaicism NIH-DHHS-US- 14-PAF07285
Co-I with Effort (Principal Investigator: Moran, John V)
05/2015-01/2020. $3,860,653 ($736,465)

5 U01 MH106892-05: 2/3 Schizophrenia Genetics and Brain Somatic Mosaicism NIH-DHHS-US- 14-PAF07285; 16-PAF05088
Co-I without Effort (Principal Investigator: Moran, John V)
05/2015-01/2020. $659,032

5 F31 NS090883-03: Upstream open reading frames in neuronal function: a singular and genome-wide approach NIH-DHHS-US- 14-PAF05593
Co-I without Effort (Principal Investigator: Rodriguez, Caitlin)
03/2015-02/2018. $100,131 ($33,377)

Submitted Grants

Discovery and analysis of structural variation in whole genome sequences NIH-DHHS-US- 17-PAF07922
Mills, Ryan Edward, PI
04/2018-03/2021. $1,397,045

1 R01 HG009896-01: Characterization and Impact of Structural Variation on Gene Regulatory Elements NIH-DHHS-US-17-PAF04387- 17-PAF04387
Mills, Ryan Edward, PI
09/2017-08/2022. $3,217,108 ($631,528)

Past Grants

4 R01 GM103961-04: Comprehensive Characterization of Canine Genomic Structural Diversity NIH-DHHS-US-12-PAF04410
Co-I with Effort (Principal Investigator: Kidd, Jeffrey)
09/2013-04/2017. $985,991 ($301,049)

Honors and Awards

National

2006-2008	Ruth L. Kirschstein National Research Service Award (NRSA) Individual Postdoctoral Fellowship
2012	Profiled in Sixth Annual Young Investigators at GenomeWeb (https://www.genomeweb.com/sequencing/ryan-mills-indels-and-lots-genomes)
2015	Highlighted in "Copy Number Analysis Starts to Add Up" in Genetic Engineering & Biotechnology news (http://www.genengnews.com/gen-articles/copy-number-analysis-starts-to-add-up/5588/)
2015	Profiled in the Journal of Young Investigators (http://www.jyi.org/issue/interview-with-a-bioinformatician-dr-ryan-mills-ph-d/)

Institutional

2014	Endowment for the Basic Sciences Teaching Award

Memberships in Professional Societies

2009-present	Member, American Society of Human Genetics
2011-present	Member, International Society for Computational Biology

Editorial Positions, Boards, and Peer-Review Service

Study Sections

National

2014	NIH/NHGRI Study Section - Interpreting Variation in Human Non-Coding Genomic Regions Using Computational Approaches and Experimental Assessment (Ad Hoc)
2015	NIH Study Section – Genomics, Computational Biology and Technology (Ad Hoc)
2015	NIH Study Section – Interpreting Variation in Human Non-Coding Genomic Regions Using Computational Approaches and Experimental Assessment (Ad Hoc)

2016	NIH Study Section – Genomics, Computational Biology and Technology (Ad Hoc)
2016	NIH Study Section – Maximizing Investigators' Research Award for New and Early Stage Investigators (Ad Hoc)

Editorial Boards

2015-present	Editorial Board Member, Scientific Reports
2017-present	Editorial Board Member, PeerJ

Journal Reviewer

2012-present	American Journal of Human Genetics (Ad Hoc)
2012-present	BMC Bioinformatics (Ad Hoc)
2012-present	Bioinformatics (Ad Hoc)
2012-present	Genome Biology (Ad Hoc)
2012-present	Genome Research (Ad Hoc)
2012-present	Nature Genetics (Ad Hoc)
2013-present	Nature Communications (Ad Hoc)
2013-present	Nature Protocols (Ad Hoc)
2014-present	Human Genetics (Ad Hoc)
2015-present	PLoS Computational Biology (Ad Hoc)
2016-present	Methods (Ad Hoc)

Teaching

Graduate Student

01/2013-06/2017	Xuefang Zhao, PhD, University of Michigan
05/2013-present	Sang (Tony) Chun, PhD, University of Michigan
01/2014-05/2014	Fan Zhang, PhD (rotation), University of Michigan
01/2014-present	Yifan Wang, PhD, University of Michigan
01/2015-present	Marcus Sherman, PhD, University of Michigan
01/2016-present	Nan Lin, MS, University of Michigan
09/2016-present	Alexandra Weber, PhD, University of Michigan
01/2017-05/2017	Catherine Barnier, PhD (rotation), University of Michigan
01/2017-present	Zhenning Zhang, MS, University of Michigan
07/2017-present	Chen Sun, PhD (rotation), University of Michigan

Postdoctoral Fellow

08/2012-07/2016	Gargi Dayama, PhD, University of Michigan
09/2015-present	Weichen Zhou, PhD, University of Michigan

Teaching Activity

National

06/2014-present	Lecturer, Mathematical and Theoretical Biology Institute, Arizona State University

Institutional

01/2012-present	PhD Candidate Preliminary Exams: Yanxiao Zhang, Brendan Veeneman, Ellen Schmidt, Andy Kong, Yindra Puentes, Shiya Song, Ridvan Eksi, Raymond Cavalcante, Laura Seaman, Alexander Kalinin, Fan Zhang, Sierra Nishizaki, Jed Carlson, Guyen Vo, Ricardo D'Oliveira Albanus, Shriya Sethuraman, Shengcheng Dong, Scott Ronquist, Peter Orchard, Christopher Castro, Ningxin Ouyang
01/2012-present	PhD Thesis Committees: Kart Tomberg, Brendan Veeneman, Killeen Kirkconnell, Xuefang Zhao, Diane Flasch, Akima George, Tony Chun, Yifan Wang, Caitlin Rodriguez, Adrian Tan, Dan Hovelson, Fan Zhang, Sierra Nishizaki, Andy Kong, Shengcheng Dong

09/2012	Lecturer, HG 632 – Experimental Genetics Systems, University of Michigan
09/2012-present	Coursemaster/Lecturer, BIOL 527 – Introduction to Bioinformatics, University of Michigan
09/2014	Lecturer, Coursera (online) – Introduction to Bioinformatics, University of Michigan
10/2014	Lecturer, UM NIEHS P30 Center and UM BRCF Bioinformatics Core Workshop – Introduction to Genome Variation
01/2015-present	Lecturer, BIOINF 525 - Foundations in Bioinformatics and Systems Biology
08/2015-present	Coursemaster/Lecturer, BIOINF/HUMGEN/BIOSTATS 606 – Introduction to Biocomputing, University of Michigan
03/2016-present	Lecturer, HUMGEN 551 – Computational Genomics, University of Michigan
03/2016-present	Lecturer, HUMGEN 803 – Current Methods, University of Michigan

Dissertation Committees

2016	Brendan Veeneman, Development and application of methods to discover cancer-associated transcript variants, University of Michigan, Computational Medicine & Bioinformatics, Committee Member
2016	Kart Tomberg, Identification of Thrombosis Modifier Genes Using ENU Mutagenesis in the Mouse, University of Michigan, Human Genetics, Committee Member
2016	Killeen Kirkconnell, Capturing transcriptional dynamics using nascent RNA sequencing, University of Michigan, Human Genetics, Committee Member
2017	Andy Kong, Computational strategies for proteogenomic analyses, University of Michigan, Computational Medicine & Bioinformatics, Committee Member
2017	Daniel H. Hovelson, Precision oncology opportunities and disease insights from next-generation sequencing profiling of routine clinical biospecimens, University of Michigan, Computational Medicine and Bioinformatics, Committee Member
2017	Xuefang Zhao, Understanding the complexity of human structural genomic variation through multiple whole genome sequencing platforms, University of Michigan, Computational Medicine & Bioinformatics, Chair

Committee and Administrative Services

Committee Services

National

2011	First RECOMB Satellite Workshop on Massively Parallel Sequencing, Program Committee, Program Committee
2015-present	Great Lakes Bioinformatics Conference, Program Committee
2015	Fifth RECOMB Satellite Workshop on Massively Parallel Sequencing, Program Committee
2016	American Society of Human Genetics, Abstract Reviewer

Institutional

2012	Bioinformatics Master's Program Admissions Commitee, Member
2013-present	Human Genetics Master's Admission Committee, Member
2014-2016	DCM&B Seminar Series Committee, Co-Chair
2014-present	Bioinformatics PhD Admissions Committee, member
2014-present	Human Genetics Picnic Committee, Chair
2016-present	DCM&B Chair's Advisory Committee, Member
2016-present	DCM&B Website Committee, Chair

Visiting Professorships and Extramural Invited Presentations

Extramural Invited Presentations

1. Improving gene annotation of cytomegalovirus genomes by statistical and comparative genomics as verified by a proteomics-based analysis of isolated MCMV virions, 9th International Cytomegalovirus Workshop, May 2003, Maastricht, Netherlands

2. Designing Custom CGH Arrays: Considerations for CNV Discovery and Genotyping, Agilent Technologies Workshop, American Society of Human Genetics, October 2009, Honolulu, HI
3. Panel Discussion: Copy Number Variants, 3rd Annual PQG Conference, Harvard School of Public Health, November 2009, Boston, MA
4. Capturing Structural Variation from Whole Genome Population-Scale Sequencing: Perspectives from the 1000 Genomes Project, Keystone Symposium on Functional Consequences of Structural Variation in the Genome, January 2011, Steamboat Springs, CO
5. Challenges in Mapping Copy Number Variation from Population-scale Genome Sequence Data, Open Science Grid, 2011 All Hands Meeting, March 2011, Boston, MA
6. Capturing Structural Variation from Whole Genome Population-Scale Sequencing: Perspectives from the 1000 Genomes Project, Cambridge Healthtech Institute, X-GEN Congress and Expo, March 2011, San Diego, CA
7. Structural Variation in the 1000 Genomes Project, Genomic Disorders 2012, Wellcome Trust Sanger Institute, March 2012, Hinxton, UK
8. Discovery and functional impact of structural variation across 1000 genomes, Cambridge Healthtech Institute, NGx: Applying Next Generation Sequencing, August 2012, Providence, RI
9. Mapping structural variation by population-scale genome sequencing, Radiation Effects Research Foundation, March 2013, Hiroshima, Japan
10. Exploring Complex Structural Genomic Variation using Next-Gen Sequencing , BioConference Live, Genetics and Genomics, August 2014, Online (Live Virtual Presentation)
11. Genomic landscape of polymorphic nuclear mitochondrial insertions in humans and other primates, American Society of Human Genetics Annual Meeting, October 2014, San Diego, CA

Other

1. 1000 Genomes Project Data Tutorial, International Congress of Human Genetics, November 2012, Montreal, Canada
2. 1000 Genomes Project Data Tutorial, American Society of Human Genetics Annual Meeting, November 2012, San Francisco, CA
3. Platform Moderator, American Society of Human Genetics Annual Meeting, November 2012, San Francisco, CA
4. Invited Session Moderator, American Society of Human Genetics Annual Meeting, October 2014, San Diego, CA

Seminars

1. Analysis of Structural Variation in the 1000 Genomes Project Pilot: New Methods, New Insights (co-speaker), Medical and Population Genetics Seminar, Broad Institute, September 2010, Boston, MA
2. Natural Structural Variation in the Human Genome, Interdisciplinary Group Seminar (IGS), Rackham Graduate School, University of Michigan, April 2012, Ann Arbor, MI
3. Navigating Genomic Complexity: Discovery and Analysis of Structural Variation, NCIBI Tools and Technology Series, University of Michigan, May 2012, Ann Arbor, MI
4. Discovery and Analysis of Structural Genomic Variation in Human Populations, School of Biology Seminar, Wabash College, October 2012, Crawfordsville, IN
5. Exploring the Hidden Genome: Deciphering Cryptic and Complex Structural Variation, Ewha Womans University, July 2016, Seoul, South Korea

Bibliography

Peer-Reviewed Journals and Publications

1. Borodovsky M, Lomsadze A, Ivanov N, Mills R: Eukaryotic gene prediction using GeneMark.hmm. Current protocols in bioinformatics / editoral board, Andreas D. Baxevanis ... [et al.] Chapter 4: Unit4.6, 2003. PM18428701
2. Perelygina L, Zhu L, Zurkuhlen H, Mills R, Borodovsky M, Hilliard JK: Complete sequence and comparative analysis of the genome of herpes B virus (Cercopithecine herpesvirus 1) from a rhesus monkey J. Virol. 77(11): 6167-6177, 2003. PM12743273

3. Mills R, Rozanov M, Lomsadze A, Tatusova T, Borodovsky M: Improving gene annotation of complete viral genomes Nucleic Acids Res. 31(23): 7041-7055, 2003. PM14627837
4. Kattenhorn LM, Mills R, Wagner M, Lomsadze A, Makeev V, Borodovsky M, Ploegh HL, Kessler BM: Identification of proteins associated with murine cytomegalovirus virions J. Virol. 78(20): 11187-11197, 2004. PM15452238
5. Mills RE, Luttig CT, Larkins CE, Beauchamp A, Tsui C, Pittard WS, Devine SE: An initial map of insertion and deletion (INDEL) variation in the human genome Genome Res. 16(9): 1182-1190, 2006. PM16902084
6. Mills RE, Bennett EA, Iskow RC, Luttig CT, Tsui C, Pittard WS, Devine SE: Recently mobilized transposons in the human and chimpanzee genomes Am. J. Hum. Genet. 78(4): 671-679, 2006. PM16532396
7. Mills RE, Bennett EA, Iskow RC, Devine SE: Which transposable elements are active in the human genome? Trends Genet. 23(4): 183-191, 2007. PM17331616
8. Lange A, Mills RE, Lange CJ, Stewart M, Devine SE, Corbett AH: Classical nuclear localization signals: Definition, function, and interaction with importin α J. Biol. Chem. 282(8): 5101-5105, 2007. PM17170104
9. Bennett EA, Keller H, Mills RE, Schmidt S, Moran JV, Weichenrieder O, Devine SE: Active Alu retrotransposons in the human genome Genome Res. 18(12): 1875-1883, 2008. PM18836035
10. Lange A, Mills RE, Devine SE, Corbett AH: A PY-NLS nuclear targeting signal is required for nuclear localization and function of the Saccharomyces cerevisiae mRNA-binding protein Hrp1 J. Biol. Chem. 283(19): 12926-12934, 2008. PM18343812
11. Borodovsky M, Mills R, Besemer J, Lomsadze A: Prokaryotic gene prediction using GeneMark and GeneMark.hmm. Curr Protoc Bioinformatics Chapter 4: Unit4.5, 2008. PM18428700
12. Kim JI, Ju YS, Park H, Kim S, Lee S, Yi JH, Mudge J, Miller NA, Hong D, Bell CJ, Kim HS, Chung IS, Lee WC, Lee JS, Seo SH, Yun JY, Woo HN, Lee H, Suh D, Lee S, Kim HJ, Yavartanoo M, Kwak M, Zheng Y, Lee MK, Park H, Kim JY, Gokcumen O, Mills RE, Zaranek AW, Thakuria J, Wu X, Kim RW, Huntley JJ, Luo S, Schroth GP, Wu TD, Kim H, Yang KS, Park WY, Kim H, Church GM, Lee C, Kingsmore SF, Seo JS: A highly annotated whole-genome sequence of a Korean individual Nature 460(7258): 1011-1015, 2009. PM19587683
13. Mullaney JM, Mills RE, Pittard WS, Devine SE: Small insertions and deletions (INDELs) in human genomes Hum. Mol. Genet. 19(R2): R131-R136, 2010. PM20858594
14. 1000 Genomes Project Consortium#, Abecasis GR, Altshuler D, Auton A, Brooks LD, Durbin RM, Gibbs RA, Hurles ME, McVean GA: A map of human genome variation from population-scale sequencing Nature 467(7319): 1061-1073, 2010. PM20981092
15. Park H, Kim JI, Ju YS, Gokcumen O, Mills RE, Kim S, Lee S, Suh D, Hong D, Kang HP, Yoo YJ, Shin JY, Kim HJ, Yavartanoo M, Chang YW, Ha JS, Chong W, Hwang GR, Darvishi K, Kim H, Yang SJ, Yang KS, Kim H, Hurles ME, Scherer SW, Carter NP, Tyler-Smith C, Lee C, Seo JS: Discovery of common Asian copy number variants using integrated high-resolution array CGH and massively parallel DNA sequencing Nat. Genet. 42(5): 400-405, 2010. PM20364138
16. Lange A, McLane LM, Mills RE, Devine SE, Corbett AH: Expanding the definition of the classical bipartite nuclear localization signal Traffic 11(3): 311-323, 2010. PM20028483
17. Iskow RC, McCabe MT, Mills RE, Torene S, Pittard WS, Neuwald AF, Van Meir EG, Vertino PM, Devine SE: Natural mutagenesis of human genomes by endogenous retrotransposons Cell 141(7): 1253-1261, 2010. PM20603005
18. Mills RE, Walter K, Stewart C, Handsaker RE, Chen K, Alkan C, Abyzov A, Yoon SC, Ye K, Cheetham RK, Chinwalla A, Conrad DF, Fu Y, Grubert F, Hajirasouliha I, Hormozdiari F, Iakoucheva LM, Iqbal Z, Kang S, Kidd JM, Konkel MK, Korn J, Khurana E, Kural D, Lam HY, Leng J, Li R, Li Y, Lin CY, Luo R, Mu XJ, Nemesh J, Peckham HE, Rausch T, Scally A, Shi X, Stromberg MP, Stütz AM, Urban AE, Walker JA, Wu J, Zhang Y, Zhang ZD, Batzer MA, Ding L, Marth GT, McVean G, Sebat J, Snyder M, Wang J, Ye K, Eichler EE, Gerstein MB, Hurles ME, Lee C, McCarroll SA, Korbel JO, 1000 Genomes Project: Mapping copy number variation by population-scale genome sequencing Nature 470(7332): 59-65, 2011. PM21293372
19. Mills RE, Pittard WS, Mullaney JM, Farooq U, Creasy TH, Mahurkar AA, Kemeza DM, Strassler DS, Ponting CP, Webber C, Devine SE: Natural genetic variation caused by small insertions and deletions in the human genome Genome Res. 21(6): 830-839, 2011. PM21460062
20. Pinto D, Darvishi K, Shi X, Rajan D, Rigler D, Fitzgerald T, Lionel AC, Thiruvahindrapuram B, Macdonald JR, Mills R, Prasad A, Noonan K, Gribble S, Prigmore E, Donahoe PK, Smith RS, Park JH, Hurles ME, Carter NP, Lee C, Scherer SW, Feuk L: Comprehensive assessment of array-based platforms and calling algorithms for detection of copy number variants Nat. Biotechnol. 29(6): 512-520, 2011. PM21552272

21. Gokcumen O, Babb PL, Iskow RC, Zhu Q, Shi X, Mills RE, Ionita-Laza I, Vallender EJ, Clark AG, Johnson WE, Lee C: Refinement of primate copy number variation hotspots identifies candidate genomic regions evolving under positive selection Genome Biol. 12(5): R52, 2011. PM21627829

22. Gravel S, Henn BM, Gutenkunst RN, Indap AR, Marth GT, Clark AG, Yu F, Gibbs RA, 1000 Genomes Project#, Bustamante CD: Demographic history and rare allele sharing among human populations. Proc Natl Acad Sci U S A 108(29): 11983-8, 2011. PM21730125/PMC3142009

23. Chen X, Shi X, Xu X, Wang Z, Mills R, Lee C, Xu J: A two-graph guided multi-task Lasso approach for eQTL mapping Proceedings of the 15th International Conference of Artificial Intelligence and Statistics (AISTATS) 22: 208-217, 2012.

24. 1000 Genomes Project Consortium#, Abecasis GR, Auton A, Brooks LD, DePristo MA, Durbin RM, Handsaker RE, Kang HM, Marth GT, McVean GA: An integrated map of genetic variation from 1,092 human genomes Nature 491(7422): 56-65, 2012. PM23128226

25. Iskow RC, Gokcumen O, Abyzov A, Malukiewicz J, Zhu Q, Sukumar AT, Pai AA, Mills RE, Habegger L, Cusanovich DA, Rubel MA, Perry GH, Gerstein M, Stone AC, Gilad Y, Lee C: Regulatory element copy number differences shape primate expression profiles Proc. Natl. Acad. Sci. U.S.A. 109(31): 12656-12661, 2012. PM22797897

26. Chiang C, Jacobsen JC, Ernst C, Hanscom C, Heilbut A, Blumenthal I, Mills RE, Kirby A, Lindgren AM, Rudiger SR, McLaughlan CJ, Bawden CS, Reid SJ, Faull RL, Snell RG, Hall IM, Shen Y, Ohsumi TK, Borowsky ML, Daly MJ, Lee C, Morton CC, MacDonald ME, Gusella JF, Talkowski ME: Complex reorganization and predominant non-homologous repair following chromosomal breakage in karyotypically balanced germline rearrangements and transgenic integration Nat. Genet. 44(4): 390-397, 2012. PM22388000

27. Brown KH, Dobrinski KP, Lee AS, Gokcumen O, Mills RE, Shi X, Chong WW, Chen JY, Yoo P, David S, Peterson SM, Raj T, Choy KW, Stranger BE, Williamson RE, Zon LI, Freeman JL, Lee C: Extensive genetic diversity and substructuring among zebrafish strains revealed through copy number variant analysis Proc. Natl. Acad. Sci. U.S.A. 109(2): 529-534, 2012. PM22203992

28. Clarke L, Zheng-Bradley X, Smith R, Kulesha E, Xiao C, Toneva I, Vaughan B, Preuss D, Leinonen R, Shumway M, Sherry S, Flicek P, 1000 Genomes Project Consortium#.: The 1000 Genomes Project: data management and community access. Nat Methods 9(5): 459-62, 2012. PM22543379/PMC3340611

29. MacArthur DG, Balasubramanian S, Frankish A, Huang N, Morris J, Walter K, Jostins L, Habegger L, Pickrell JK, Montgomery SB, Albers CA, Zhang ZD, Conrad DF, Lunter G, Zheng H, Ayub Q, DePristo MA, Banks E, Hu M, Handsaker RE, Rosenfeld JA, Fromer M, Jin M, Mu XJ, Khurana E, Ye K, Kay M, Saunders GI, Suner MM, Hunt T, Barnes IH, Amid C, Carvalho-Silva DR, Bignell AH, Snow C, Yngvadottir B, Bumpstead S, Cooper DN, Xue Y, Romero IG, 1000 Genomes Project Consortium#, Wang J, Li Y, Gibbs RA, McCarroll SA, Dermitzakis ET, Pritchard JK, Barrett JC, Harrow J, Hurles ME, Gerstein MB, Tyler-Smith C: A systematic survey of loss-of-function variants in human protein-coding genes. Science 335(6070): 823-8, 2012. PM22344438/PMC3299548

30. Xue Y, Chen Y, Ayub Q, Huang N, Ball EV, Mort M, Phillips AD, Shaw K, Stenson PD, Cooper DN, Tyler-Smith C, 1000 Genomes Project Consortium#.: Deleterious- and disease-allele prevalence in healthy individuals: insights from current predictions, mutation databases, and population-scale resequencing. Am J Hum Genet 91(6): 1022-32, 2012. PM23217326/PMC3516590

31. Rogers AJ, Chu JH, Darvishi K, Ionita-Laza I, Lehmann H, Mills R, Lee C, Raby BA: Copy number variation prevalence in known asthma genes and their impact on asthma susceptibility. Clinical and experimental allergy : journal of the British Society for Allergy and Clinical Immunology 43(4): 455-62, 2013. PM23517041

32. Silva AG, Krepischi AC, Torrezan GT, Capelli LP, Carraro DM, D'Angelo CS, Koiffmann CP, Zatz M, Naslavsky MS, Masotti C, Otto PA, Achatz MI, Mills RE, Lee C, Pearson PL, Rosenberg C: Does germ-line deletion of the PIP gene constitute a widespread risk for cancer? European journal of human genetics : EJHG 22(3): 307, 2013. PM23778870

33. Montgomery SB, Goode DL, Kvikstad E, Albers CA, Zhang ZD, Mu XJ, Ananda G, Howie B, Karczewski KJ, Smith KS, Anaya V, Richardson R, Davis J, 1000 Genomes Project Consortium#, MacArthur DG, Sidow A, Duret L, Gerstein M, Makova KD, Marchini J, McVean G, Lunter G: The origin, evolution, and functional impact of short insertion-deletion variants identified in 179 human genomes. Genome Res 23(5): 749-61, 2013. PM23478400/PMC3638132

34. Khurana E, Fu Y, Colonna V, Mu XJ, Kang HM, Lappalainen T, Sboner A, Lochovsky L, Chen J, Harmanci A, Das J, Abyzov A, Balasubramanian S, Beal K, Chakravarty D, Challis D, Chen Y, Clarke D, Clarke L, Cunningham F, Evani US, Flicek P, Fragoza R, Garrison E, Gibbs R, Gümüs ZH, Herrero J, Kitabayashi N, Kong Y, Lage K, Liluashvili V, Lipkin SM, MacArthur DG, Marth G, Muzny D, Pers TH, Ritchie GR, Rosenfeld JA, Sisu C, Wei X, Wilson M, Xue Y, Yu F, 1000 Genomes Project Consortium#, Dermitzakis ET, Yu H, Rubin MA, Tyler-Smith C, Gerstein M: Integrative annotation of variants from 1092 humans: application to cancer genomics. Science 342(6154): 1235587, 2013. PM24092746/PMC3947637

35. Abyzov A, Iskow R, Gokcumen O, Radke DW, Balasubramanian S, Pei B, Habegger L, 1000 Genomes Project Consortium#, Lee C, Gerstein M: Analysis of variable retroduplications in human populations suggests coupling of retrotransposition to cell division. Genome Res 23(12): 2042-52, 2013. PM24026178/PMC3847774

36. Todd PK, Oh SY, Krans A, He F, Sellier C, Frazer M, Renoux AJ, Chen KC, Scaglione KM, Basrur V, Elenitoba-Johnson K, Vonsattel JP, Louis ED, Sutton MA, Taylor JP, Mills RE, Charlet-Berguerand N, Paulson HL: CGG repeat-associated translation mediates neurodegeneration in fragile X tremor ataxia syndrome Neuron 78(3): 440-455, 2013. PM23602499

37. Chu JH, Rogers A, Ionita-Laza I, Darvishi K, Mills RE, Lee C, Raby BA: Copy number variation genotyping using family information. BMC bioinformatics 14: 157, 2013. PM23656838

38. Gokcumen O, Tischler V, Tica J, Zhu Q, Iskow RC, Lee E, Fritz MH, Langdon A, Stütz AM, Pavlidis P, Benes V, Mills RE, Park PJ, Lee C, Korbel JO: Primate genome architecture influences structural variation mechanisms and functional consequences. Proceedings of the National Academy of Sciences of the United States of America 110(39): 15764-9, 2013. PM24014587

39. Park H, Kim D, Kim CH, Mills RE, Chang MY, Iskow RC, Ko S, Moon JI, Choi HW, Man Yoo PS, Do JT, Han MJ, Lee EG, Jung JK, Zhang C, Lanza R, Kim KS: Increased genomic integrity of an improved protein-based mouse induced pluripotent stem cell method compared with current viral-induced strategies. Stem cells translational medicine 3(5): 599-609, 2014. PM24763686

40. Brand H, Pillalamarri V, Collins RL, Eggert S, O'Dushlaine C, Braaten EB, Stone MR, Chambert K, Doty ND, Hanscom C, Rosenfeld JA, Ditmars H, Blais J, Mills R, Lee C, Gusella JF, McCarroll S, Smoller JW, Talkowski ME, Doyle AE: Cryptic and complex chromosomal aberrations in early-onset neuropsychiatric disorders. American journal of human genetics 95(4): 454-61, 2014. PM25279985

41. Dayama G, Emery SB, Kidd JM, Mills RE: The genomic landscape of polymorphic human nuclear mitochondrial insertions Nucleic Acids Res. 42(20): 12640-12649, 2014. PM25348406

42. Delaneau O, Marchini J, 1000 Genomes Project Consortium#.: Integrating sequence and array data to create an improved 1000 Genomes Project haplotype reference panel. Nature Communications 5: 3934, 2014. PM25653097

43. Colonna V, Ayub Q, Chen Y, Pagani L, Luisi P, Pybus M, Garrison E, Xue Y, Tyler-Smith C, 1000 Genomes Project Consortium#, Abecasis GR, Auton A, Brooks LD, DePristo MA, Durbin RM, Handsaker RE, Kang HM, Marth GT, McVean GA: Human genomic regions with exceptionally high levels of population differentiation identified from 911 whole-genome sequences. Genome Biol 15(6): R88, 2014. PM24980144/PMC4197830

44. 1000 Genomes Project Consortium#, Auton A, Brooks LD, Durbin RM, Garrison EP, Kang HM, Korbel JO, Marchini JL, McCarthy S, McVean GA, Abecasis GR: A global reference for human genetic variation Nature 526(7571): 68-74, 2015. PM26432245

45. Sudmant PH, Rausch T, Gardner EJ, Handsaker RE, Abyzov A, Huddleston J, Zhang Y, Ye K, Jun G, Hsi-Yang Fritz M, Konkel MK, Malhotra A, Stütz AM, Shi X, Paolo Casale F, Chen J, Hormozdiari F, Dayama G, Chen K, Malig M, Chaisson MJ, Walter K, Meiers S, Kashin S, Garrison E, Auton A, Lam HY, Jasmine Mu X, Alkan C, Antaki D, Bae T, Cerveira E, Chines P, Chong Z, Clarke L, Dal E, Ding L, Emery S, Fan X, Gujral M, Kahveci F, Kidd JM, Kong Y, Lameijer EW, McCarthy S, Flicek P, Gibbs RA, Marth G, Mason CE, Menelaou A, Muzny DM, Nelson BJ, Noor A, Parrish NF, Pendleton M, Quitadamo A, Raeder B, Schadt EE, Romanovitch M, Schlattl A, Sebra R, Shabalin AA, Untergasser A, Walker JA, Wang M, Yu F, Zhang C, Zhang J, Zheng-Bradley X, Zhou W, Zichner T, Sebat J, Batzer MA, McCarroll SA, 1000 Genomes Project Consortium, Mills RE*, Gerstein MB*, Bashir A*, Stegle O*, Devine SE*, Lee C*, Eichler EE*, Korbel JO*: An integrated map of structural variation in 2,504 human genomes. Nature 526(7571): 75-81, 2015. PM26432246

46. Jorge DM, Mills RE, Lauring AS: CodonShuffle: a tool for generating and analyzing synonymously mutated sequences. Virus Evol 1(1): vev012, 2015. PM27774284/PMC5014483

47. Chun SY, Rodriguez CM, Todd PK, Mills RE: SPECtre: a spectral coherence--based classifier of actively translated transcripts from ribosome profiling sequence data. BMC Bioinformatics 17(1): 482, 2016. PM27884106/PMC5123373

48. Fang Q, George AS, Brinkmeier ML, Mortensen AH, Gergics P, Cheung LY, Daly AZ, Ajmal A, Pérez Millán MI, Ozel AB, Kitzman JO, Mills RE, Li JZ, Camper SA: Genetics of Combined Pituitary Hormone Deficiency: Roadmap into the Genome Era. Endocr Rev 37(6): 636-675, 2016. PM27828722/PMC5155665

49. Zhao X, Emery SB, Myers B, Kidd JM, Mills RE: Resolving complex structural genomic rearrangements using a randomized approach. Genome Biol 17(1): 126, 2016. PM27287201/PMC4901421

50. Zhao X, Weber AM, Mills RE: A recurrence-based approach for validating structural variation using long-read sequencing technology. Gigascience 6(8): 1-9, 2017. PM28873962

51. McConnell MJ, Moran JV, Abyzov A, Akbarian S, Bae T, Cortes-Ciriano I, Erwin JA, Fasching L, Flasch DA, Freed D, Ganz J, Jaffe AE, Kwan KY, Kwon M, Lodato MA, Mills RE, Paquola ACM, Rodin RE, Rosenbluh C, Sestan N, Sherman MA, Shin JH, Song S, Straub RE, Thorpe J, Weinberger DR, Urban AE, Zhou B, Gage FH, Lehner T, Senthil G, Walsh CA, Chess A, Courchesne E, Gleeson JG, Kidd JM, Park PJ, Pevsner J, Vaccarino FM, Brain Somatic Mosaicism Network.: Intersection of diverse neuronal genomes and neuropsychiatric disease: The Brain Somatic Mosaicism Network. Science 356(6336): 2017. PM28450582

52. Gardner EJ, Lam VK, Harris DN, Chuang NT, Scott EC, Pittard WS, Mills RE, 1000 Genomes Project Consortium., Devine SE: The Mobile Element Locator Tool (MELT): Population-scale mobile element discovery and biology. Genome Res: 2017. PM28855259

53. Hovelson DH, Liu C, Wang Y, Kang Q, Henderson J, Gursky A, Ramnath N, Krauss JC, Talpaz M, Kandarpa M, Chugh R, Tuck M, Herman K, Grasso CS, Quist MJ, Feng FY, Haakenson C, Langmore J, Kamberov E, Tesmer T, Husain H, Lonigro RJ, Robinson D, Smith DC, Alva AS, Hussain MH, Chinnaiyan AM, Tewari M, Mills RE, Morgan, Tomlins SA: Rapid, Ultra Low Coverage Copy Number Profiling of Cell-Free DNA as a Precision Oncology Screening Strategy Oncotarget: 2017. (In Press)

Non-Peer-Reviewed Journals and Publications

Submitted

1. Chaisson MJP, Sanders AD, Zhao X, Malhotra A, Porubsky D, Rausch T, Gardner EJ, Rodriguez O, Guo L, Collins RL, Fan X, Wen J, Handsaker RE, Fairley S, Kronenberg ZN, Kong X, Hormozdiari F, Lee D, Wenger AM, Hastie A, Antaki D, Audano P, Brand H, Cantsilieris S, Cao H, Cerveira E, Chen C, Chen X, Chin C, Chong Z, Chuang NT, Church DM, Clarke L, Farrell A, Flores J, Galeev T, David G, Gujral M, Guryev V, Haynes-Heaton W, Korlach J, Kumar S, Kwon JY, Lee JE, Lee J, Lee W, Lee SP, Marks P, Valud-Martinez K, Meiers S, Munson KM, Navarro F, Nelson BJ, Nodzak C, Noor A, Kyriazopoulou-Panagiotopoulou S, Pang A, Qiu Y, Rosanio G, Ryan M, Stutz A, Spierings DCJ, Ward A, Welsch AE, Xiao M, Xu W, Zhang C, Zhu Q, Zheng-Bradley X, Jun G, Ding L, Koh CL, Ren B, Flicek P, Chen K, Gerstein MB, Kwok P, Lansdorp PM, Marth G, Sebat J, Shi X, Bashir A, Ye K, Devine SE, Talkowski M, Mills RE, Marschall T, Korbel JO, Eichler EE, Lee C: Multi-platform discovery of haplotype-resolved structural variation in human genomes BioRxiv doi: https://doi.org/10.1101/193. (Submitted)