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@umich.edu

Education and Training

Education

08/1996-05/2000	AB, 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 and Administrative 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, Computational Medicine & Bioinformatics, University of Michigan, Ann Arbor, MI
01/2012-present	Assistant Professor, Human Genetics, University of Michigan, 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

Grants

Current Grants

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
09/2013-08/2016. $397,471 ($94,527)

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

5 R01 HG007068-04: Discovery and analysis of structural variation in whole genome sequences
NIH-DHHS-US- 13-PAF00068
Mills, Ryan Edward, PI
09/2013-07/2017. $1,526,575 ($382,699)

2/3 Schizophrenia Genetics and Brain Somatic Mosaicism
NIH-DHHS-US- 14-PAF07285
Moran, John V, Co-I with Effort
04/2015-03/2020. $4,998,681

Unconventional translation in Neuronal function and Neurologic Disease
Todd, Peter K, Co-PI with effort
05/2014-04/2016. $150,000 ($150,000)

Fidelity, robustness, and diversity in RNA virus evolution and pathogenesis
NIH-DHHS-US- 15-PAF00792
Lauring, Adam, Co-I with Effort
07/2015-06/2020. $2,134,000

Honors and Awards

National

2012	GenomeWeb: Genome Technology "Young Investigator"

Institutional

2014	Endowment for the Basic Sciences Teaching Award

Memberships in Professional Societies

2009-present	Member, American Society of Human Genetics
2001-present	Member, International Society for Computational Biology

Editorial Positions, Boards, and Peer-Review Service

Editorial Boards

2015-present	Scientific Reports (Board Member)

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)

Study Sections

National

2014	NIH Study Section – Interpreting Variation in Human Non-Coding Genomic Regions Using Computational Approaches and Experimental Assessment (Ad Hoc)
2015	NIH 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)
2016	NIH Study Section – Maximizing Investigators' Research Award for New and Early Stage Investigators (Ad Hoc)

Teaching

Graduate Student

01/2013-present	Xuefang Zhao, BS, University of Michigan
05/2013-present	Sang (Tony) Chun, MS, University of Michigan
01/2014-present	Yifan Wang, BS, University of Michigan
01/2014	Fan Zhang, BS, University of Michigan (rotation)
09/2014-present	Akima George, BS, University of Michigan (co-mentor)
01/2015-present	Marcus Sherman, BS, University of Michigan (rotation)

Postdoctoral Fellow

08/2012-present	Gargi Dayama, PhD, University of Michigan
09/2015-present	Weichen (Arthur) Zhou, University of Michigan

Teaching Activity

National

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

Institutional

09/2012	Lecturer, HUMGEN 632 – Experimental Genetics Systems, University of Michigan
09/2012-present	Coursemaster/Lecturer, BIOINF 527 – Introduction to Bioinformatics, University of Michigan
01/2015-present	Lecturer, BIOINF 525 - Foundations in Bioinformatics and Systems Biology
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

08/2015-present	Coursemaster/Lecturer, BIOINF/HUMGEN/BIOSTATS 606 – Introduction to Biocomputing, University of Michigan
03/2016	Lecturer, HUMGEN 803 – Current Methods, University of Michigan
03/2016	Lecturer, HUMGEN 551 – Computational Genomics, University of Michigan

Committee and Administrative Services

Committee Services

National

2011	First RECOMB Satellite Workshop on Massively Parallel Sequencing, Program Committee
2015-2016	Great Lakes Bioinformatics Conference, Program Committee
2015	Fifth RECOMB Satellite Workshop on Massively Parallel Sequencing, Program Committee

Institutional

2012	Bioinformatics Master's Program Admissions Committee, Member
2013	Human Genetics Master's Program Assessment Ad-hoc Committee, Member
2014-present	Bioinformatics Website Committee, Member
2014-present	Bioinformatics Seminar Series Committee, Co-Chair
2014-present	Bioinformatics Graduate Admissions Committee, Member
2014-present	Human Genetics Picnic Committee, Co-Chair
2015-present	Human Genetics Master's Program Admissions Committee, Member

Visiting Professorships and Extramural 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, American Society of Human Genetics Annual Meeting, November 2012, San Francisco, CA
2. Platform Moderator, American Society of Human Genetics Annual Meeting, November 2012, San Francisco, CA
3. 1000 Genomes Project Data Tutorial, International Congress of Human Genetics, November 2012, Montreal, Canada
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

Media

January 2015 – Profiled in the Journal of Young Investigators (http://www.jyi.org/issue/interview-with-a-bioinformatician-dr-ryan-mills-ph-d/)

October 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/)

Bibliography

Non-Peer Reviewed Journals and Publications

1. SY Chun, CM Rodriguez, PK Todd, RE Mills. SPECtre: a spectral coherence-based classifier of actively translated transcripts from ribosome profiling sequence data. bioRxiv, 2015, DOI: 10.1101/034777

Peer-Reviewed Journals and Publications

1. 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. 2003 Jun;77(11):6167-77. PMC155011
2. Borodovsky M, **Mills R**, Besemer J, Lomsadze A. Prokaryotic gene prediction using GeneMark and GeneMark.hmm. Curr Protoc Bioinformatics. 2003 May;Chapter :Unit4.5.
3. Borodovsky M, Lomsadze A, Ivanov N, **Mills R**. Eukaryotic gene prediction using GeneMark.hmm. Curr Protoc Bioinformatics. 2003 May;Chapter 4:Unit4.6.
4. **Mills R**, Rozanov M, Lomsadze A, Tatusova T, Borodovsky M. Improving gene annotation of complete viral genomes. Nucleic Acids Res. 2003 Dec 1;31(23):7041-55. PMC290248
5. 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. 2004 Oct;78(20):11187-97. PMC521832
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. 2006 Apr;78(4):671-9. PMC142469
7. **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. 2006 Sep;16(9):1182-90. PMC1557762
8. Lange A, **Mills RE**, Lange CJ, Stewart M, Devine SE, Corbett AH. Classical nuclear localization signals: definition, function, and interaction with importin alpha. J Biol Chem. 2007 Feb 23;282(8):5101-5.
9. **Mills RE**, Bennett EA, Iskow RC, Devine SE. Which transposable elements are active in the human genome? Trends Genet. 2007 Apr;23(4):183-91.
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. 2008 May 9;283(19):12926-34. PMC2442336
11. Bennett EA, Keller H, **Mills RE**, Schmidt S, Moran JV, Weichenrieder O, Devine SE. Active Alu retrotransposons in the human genome. Genome Res. 2008 Dec;18(12):1875-83. PMC2593586
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. 2009 Aug 20;460(7258):1011-5. PMC2860965
13. Lange A, McLane LM, **Mills RE**, Devine SE, Corbett AH. Expanding the definition of the classical bipartite nuclear localization signal. Traffic. 2010 Mar;11(3):311-23. PMC2886731

14. 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. 2010 May;42(5):400-5. PMC3329635

15. 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. 2010 Jun 25;141(7):1253-61. PMC2943760

16. Mullaney JM, **Mills RE**, Pittard WS, Devine SE. Small insertions and deletions (INDELs) in human genomes. Hum Mol Genet. 2010 Oct 15;19(R2):R131-6. PMC2953750

17. **1000 Genomes Project Consortium.** A map of human genome variation from population-scale sequencing. Nature. 2010 Oct 28;467(7319):1061-73. PMC3042601

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. 2011 Feb 3;470(7332):59-65. PMC3077050

19. 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. 2011 May 8;29(6):512-20. PMC3270583

20. **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. 2011 Jun;21(6):830-9. PMC3106316

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. 2011 May 31;12(5):R52. PMC3219974

22. 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. 2012 Jan 10;109(2):529-34. PMC3258620

23. 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 RLM, Snell RG, Hall IM, Shen Y, Ohsumi TK, Borowsky ML, Daly MJ, Lee C, Morton CC, MacDonald ME, Gusella JF, Talkowski ME. Complex genomic reorganization and a predominance of non-homologous repair following chromosomal breakage in balanced germline rearrangements and transgenic integration. Nat Genet. 2012 Mar 4;44(4):390-7. PMC3340016

24. 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. 2012 Jul 31;109(31):12656-61 PMC3411951

25. Chen X, Shi X, Xu X, Wang Z, **Mills RE**, Lee C, and Xu J. A two-graph guided multi-task lasso approach for eQTL mapping. In *Proceedings of the 15th International Conference of Artificial Intelligence and Statistics (AISTATS) 2012*, La Palma, Canary Islands, JMLR W&CP 22: 208-217

26. **1000 Genomes Project Consortium.** An integrated map of genetic variation from 1,092 human genomes. Nature. 2012 Nov 1;491(7422):56-65 PMC3498066

27. Rogers A, Chu JH, Darvishi K, Ionita-Laza I, Lehmann H, **Mills RE**, Lee C, Raby BA. Copy number variation prevalence in known asthma genes and their impact of asthma susceptibility. Clin Exp Allergy, 2013 Apr;43(4):455-62. PMC3609036

28. Todd PK, Oh S, Krans A, He F, Sellier S, Frazer M, Renoux AJ, Chen KC, Scaglione KM, Basrur V, Elenitoba-Johnson K, Vonsattel JP, Louis ED, Sutton MA, Taylor JP, **Mills RE**, Charlet-Bergurand N, Paulson HL. CGG repeat associated translation mediates neurodegeneration in Fragile X-assocated tremor ataxia syndrome. Neuron, 2013 May 8;78(3):440-55 PMC3831531

29. Chu J, Rogers AJ, Ionita-Laza I, Darvishi K, **Mills RE**, Lee C, Raby BA. Copy Number Variation Genotyping Using Family Information. BMC Bioinformatics, 2013 May 9;14:157. PMC3668900

30. 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? Eur J Hum Genet., 2014 Mar;22(3):307-9 PMC3925262

31. Gokcumen O, Tischler V, Tica J, Zhu Q, Iskow R, Lee E, Fritz MHF, Langdon A, Stutz AM, Pavlidis P, Benes V, **Mills RE**, Park PJ, Lee C, Korbel JO. Primate genome architecture influences structural variation mechanisms and functional consequences. Proc Natl Acad Sci U S A. 2013, 2013 Sep 24;110(39):15764-9 PMC3785719

32. 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. PMC24763686

33. 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. Am J Hum Genet. 2014 Oct;95(4):454-461. PMC4185111

34. Dayama G, Emery SB, Kidd JM, **Mills RE**. The genomic landscape of polymorphic human nuclear mitochondrial insertions. *Nucleic Acids Res*. 2014 Nov 10;42(20):12640-9. PMC4227756

35. Sudmant PH, Rausch T, Gardner EJ, Handsaker RE, Abyzov A, Huddleston J, Zhang Y, Ye K, Jun G, Fritz MH, Konkel MK, Malhotra A, Stutz AM, Shi X, Casale FP, Chen J, Hormozdiari F, Dayama G, Chen K, Malig M, Chaisson MJP, Walter K, Meiers S, Kashin S, Garrison E, Auton A, Lam HYK, Mu XJ, 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, Kon Y, Lameijer EW, McCarthy S, Flicek P, Gibbs RA, Marth G, Menelaou A, Muzny DM,

Neslon BJ, Noor A, Parrish NF, Quitadamo A, Raeder B, Schadt E, 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, The 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 2,504 human genomes. Nature. 2015 Oct 1;526(7571):75-81. *(* co-senior author) PMC4617611*

36. 1000 Genomes Project Consortium. A global reference for human genetic variation. Nature. 2015 Oct 1;526(7571):68-74 PMC4750478

37. Jorge DM, **Mills RE**, Lauring AS. CodonShuffle: A tool for generating and analyzing synonymously mutated sequences. Virus Evolution, 2015, 1(1): vev012

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

Peer-Reviewed Consortium Publications

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

2. 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. 2012;335(6070):823-8. PMC3299548

3. 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. 2013;23(5):749-61. PMC3638132

4. 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. 2012;91(6):1022-32. PMC3516590

5. 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. 2012;9(5):459-62. PMC3340611

6. 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. 2013;23(12):2042-52. PMC3847774

7. 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, Gumus 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. 2013;342(6154):1235587. PMC3947637

8. 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. 2014;15(6):R88. PMC4197830

9. Delaneau O, Marchini J; 1000 Genomes Project Consortium; 1000 Genomes Project Consortium. Integrating sequence and array data to create an improved 1000 Genomes Project haplotype reference panel. Nat Commun. 2014 Jun 13;5:3934. PMC4338501