Ryan E Mills, Ph.D.
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Associate Professor, Department of Human Genetics
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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
01/2012-08/2018	Assistant Professor, Human Genetics, University of Michigan - Ann Arbor, Ann Arbor, MI
01/2012-08/2018	Assistant Professor, Computational Medicine & Bioinformatics, University of Michigan - Ann Arbor, Ann Arbor, MI
09/2018-present	Associate Professor, Human Genetics, University of Michigan - Ann Arbor, Ann Arbor, MI
09/2018-present	Associate Professor with Tenure, 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 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

Pending Grants

Comprehensive Detection of Haplotype-Resolved Structural Variation in Human Genomes NIH-DHHS-US through a consortium with Jackson Laboratory- 18-PAF07010
Mills, Ryan Edward, PI-on-Sub
04/2019-03/2023. $635,600

Submitted Grants

Characterization and Impact of Structural Variation on Gene Regulatory Elements NIH-DHHS-US- 18-PAF03689
Boyle, Alan P;Mills, Ryan Edward, PI
09/2018-08/2023. $1,880,010

Discovery and analysis of structural variation in whole genome sequences NIH-DHHS-US- 18-PAF04355
Mills, Ryan Edward, PI
09/2018-08/2021. $1,182,641

Evolutionary Trajectories of Cryptic Genomic Structural Variants in Primates NIH-DHHS-US- PAR-17-482
Co-I with Effort (Principal Investigator: Gokcumen, Omer)
07/2019-06/2023. $250,000

Past 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
09/2013-08/2018. $397,471 ($94,527)

An Integrative Analysis of Structural Variation for the 1000 Genomes Project SubK-NIH-DHHS-US through a consortium with Jackson Laboratory- 18-PAF01525
Mills, Ryan Edward, PI
09/2017-08/2018. $84,451 ($84,451)

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)

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)

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 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)

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/)
2018	Profiled in Georgia Tech College of Sciences (https://cos.gatech.edu/homecoming/2018-mills)

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)
2018	NIH Study Section - Rare Genetic Disorders as a Window into the Genetic Architecture of Mental Disorders (Co-Chair)
2019	NIH Study Section – Genomics, Computational Biology and Technology (Ad Hoc)

Editorial Boards

2015-2019	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)
2012-present	Nature Methods (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)
2015-present	Nucleic Acids Research (Ad Hoc)
2016-present	Methods (Ad Hoc)
2018-present	European Journal of Human Genetics (Ad Hoc)

Teaching

Undergraduate Student

01/2019-present	Samantha Rondeau, University of Michigan

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
01/2014-05/2019	Yifan Wang, PhD, University of Michigan
01/2015-present	Marcus Sherman, PhD, University of Michigan
01/2016-12/2016	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
01/2017-12/2017	Zhenning Zhang, MS, University of Michigan
07/2017-present	Chen Sun, PhD, University of Michigan
11/2018-present	Wenjin Gu, MS, University of Michigan
01/2019-present	Steve Ho, PhD, University of Michigan

Postdoctoral Fellow

08/2012-07/2016	Gargi Dayama, PhD, University of Michigan
09/2015-present	Weichen Zhou, PhD, University of Michigan
06/2019-present	Yifan Wang, 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, Marlena Duda, Kevin Hu
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, Marcus Sherman, Scott Ronquist, Hillary Miller, Chris Castro, Kevin Hu, James Delorme, Ningxin Ouyang
09/2012	Lecturer, HG 632 – Experimental Genetics Systems, University of Michigan
09/2012-09/2018	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
01/2019-present	Coursemaster/Lecturer, BIOINF 529 - Bioinformatics Concepts and Algorithms, 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
2017	Sang Chun, Development and Application of Next-Generation Sequencing Methods to Profile Cellular Translational Dynamics, University of Michigan, Computational Medicine & Bioinformatics, Chair
2018	Caitlin Rodriguez, The role of upstream open reading frames in regulating neuronal protein synthesis, University of Michigan, Neuroscience, Committee Member
2019	Fan Zhang, Leveraging Genetic Variants for Rapid, Robust, and Scalable Analysis of Massive Sequence Datasets, University of Michigan, Computational Medicine and Bioinformatics, Committee Member
2019	Yifan Wang, Detection of Rare Events in Complex Sequencing Data, University of Michigan, Computational Medicine and 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
2018	5th Intl. Conference on Algorithms for Computational Biology, Program Committee
2018	American Society of Human Genetics, Abstract Reviewer
2019-present	National Association of Wabash Men, Wabash College, Board of Directors

Institutional

2012	Bioinformatics Master's Program Admissions Committee, Member
2014-2016	DCM&B Seminar Series Committee, Co-Chair
2014-2017	Human Genetics Picnic Committee, Chair
2013-present	Human Genetics Master's Admission Committee, Member
2014-present	Bioinformatics PhD Admissions Committee, member
2016-present	DCM&B Chair's Advisory Committee, Member
2016-present	DCM&B Website Committee, Chair
2017-present	Human Genetics Communications Committee, Member
2017-present	EBS IT committee, Medical School, Member
2018-present	HITS Learning Services Governance Committee, Medical School, Member
2019-present	Human Genetics Chair’s Advisory Committee, Member
2019-present	PIBS Curriculum Committee, Medical School, Member

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
12. Exploring the Hidden Genome: Deciphering Cryptic and Complex Structural Variation, Ewha Womans University, July 2016, Seoul, South Korea
13. Excavating the Deep Genome: Deciphering Structural Variation in Complex and Repetitive Regions. Department of Biological Sciences, Oakland University, April 2019, Rochester, MI

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. Excavating the Deep Genome: Deciphering Structural Variation in Complex and Repetitive Regions, Basic Science Seminar, Internal Medicine Department, Nephrology Division, University of Michigan, April 2018, Ann Arbor, MI
6. Exploring the dynamics of protein translation in a model of neuronal differentiation. RNA Innovation Seminar Series, Center for RNA Biomedicine, University of Michigan, May 2018, Ann Arbor, MI
7. Overview of Research Projects, Lee Lab Reunion and Symposium, The Jackson Lab for Genomic Medicine, October 2018, Farmington, CT

Bibliography

Peer-Reviewed Journals and Publications

1. Borodovsky M, Lomsadze A, Ivanov N, Mills R. Eukaryotic gene prediction using GeneMark.hmm. Curr Protoc Bioinformatics. 2003;Chapter 4:Unit4.6. Epub 2008/04/23. doi: 10.1002/0471250953.bi0406s01. PubMed PMID: 18428701.

2. Borodovsky M, Mills R, Besemer J, Lomsadze A. Prokaryotic gene prediction using GeneMark and GeneMark.hmm. Curr Protoc Bioinformatics. 2003;Chapter 4:Unit4.5. Epub 2008/04/23. doi: 10.1002/0471250953.bi0405s01. PubMed PMID: 18428700.

3. Mills R, Rozanov M, Lomsadze A, Tatusova T, Borodovsky M. Improving gene annotation of complete viral

genomes. Nucleic Acids Res. 2003;31(23):7041-55. Epub 2003/11/25. PubMed PMID: 14627837; PMCID: PMC290248.

4. 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;77(11):6167-77. Epub 2003/05/14. PubMed PMID: 12743273; PMCID: PMC155011.

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;78(20):11187-97. Epub 2004/09/29. doi: 10.1128/jvi.78.20.11187-11197.2004. PubMed PMID: 15452238; PMCID: 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;78(4):671-9. Epub 2006/03/15. doi: 10.1086/501028. PubMed PMID: 16532396; PMCID: PMC1424692.

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;16(9):1182-90. Epub 2006/08/12. doi: 10.1101/gr.4565806. PubMed PMID: 16902084; PMCID: 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;282(8):5101-5. Epub 2006/12/16. doi: 10.1074/jbc.R600026200. PubMed PMID: 17170104; PMCID: PMC4502416.

9. Mills RE, Bennett EA, Iskow RC, Devine SE. Which transposable elements are active in the human genome? Trends Genet. 2007;23(4):183-91. Epub 2007/03/03. doi: 10.1016/j.tig.2007.02.006. PubMed PMID: 17331616.

10. Bennett EA, Keller H, Mills RE, Schmidt S, Moran JV, Weichenrieder O, Devine SE. Active Alu retrotransposons in the human genome. Genome Res. 2008;18(12):1875-83. Epub 2008/10/07. doi: 10.1101/gr.081737.108. PubMed PMID: 18836035; PMCID: PMC2593586.

11. 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;283(19):12926-34. Epub 2008/03/18. doi: 10.1074/jbc.M800898200. PubMed PMID: 18343812; PMCID: PMC2442336.

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;460(7258):1011-5. Epub 2009/07/10. doi: 10.1038/nature08211. PubMed PMID: 19587683; PMCID: PMC2860965.

13. 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. 2010;467(7319):1061-73. Epub 2010/10/29. doi: 10.1038/nature09534. PubMed PMID: 20981092; PMCID: PMC3042601.

14. 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;141(7):1253-61. Epub 2010/07/07. doi: 10.1016/j.cell.2010.05.020. PubMed PMID: 20603005; PMCID: PMC2943760.

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;141(7):1253-61. Epub 2010/07/07. doi: 10.1016/j.cell.2010.05.020. PubMed PMID: 20603005; PMCID: PMC2943760.

16. Lange A, McLane LM, Mills RE, Devine SE, Corbett AH. Expanding the definition of the classical bipartite nuclear localization signal. Traffic. 2010;11(3):311-23. Epub 2009/12/24. doi: 10.1111/j.1600-0854.2009.01028.x. PubMed PMID: 20028483; PMCID: PMC2886731.

17. Mullaney JM, Mills RE, Pittard WS, Devine SE. Small insertions and deletions (INDELs) in human genomes. Hum Mol Genet. 2010;19(R2):R131-6. Epub 2010/09/23. doi: 10.1093/hmg/ddq400. PubMed PMID: 20858594; PMCID: PMC2953750.

18. 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;42(5):400-5. Epub 2010/04/07. doi: 10.1038/ng.555. PubMed PMID: 20364138; PMCID: PMC3329635.

19. Sudmant PH, Kitzman JO, Antonacci F, Alkan C, Malig M, Tsalenko A, Sampas N, Bruhn L, Shendure J, Eichler EE. Diversity of human copy number variation and multicopy genes. Science. 2010;330(6004):641-6. Epub 2010/10/30. doi: 10.1126/science.1197005. PubMed PMID: 21030649; PMCID: PMC3020103.

20. Conrad DF, Keebler JE, DePristo MA, Lindsay SJ, Zhang Y, Casals F, Idaghdour Y, Hartl CL, Torroja C, Garimella KV, Zilversmit M, Cartwright R, Rouleau GA, Daly M, Stone EA, Hurles ME, Awadalla P. Variation in genome-wide mutation rates within and between human families. Nat Genet. 2011;43(7):712-4. Epub 2011/06/15. doi: 10.1038/ng.862. PubMed PMID: 21666693; PMCID: PMC3322360.

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;12(5):R52. Epub 2011/06/02. doi: 10.1186/gb-2011-12-5-r52. PubMed PMID: 21627829; PMCID: PMC3219974.

22. Gravel S, Henn BM, Gutenkunst RN, Indap AR, Marth GT, Clark AG, Yu F, Gibbs RA, Bustamante CD. Demographic history and rare allele sharing among human populations. Proc Natl Acad Sci U S A. 2011;108(29):11983-8. Epub 2011/07/07. doi: 10.1073/pnas.1019276108. PubMed PMID: 21730125; PMCID: PMC3142009.

23. Marth GT, Yu F, Indap AR, Garimella K, Gravel S, Leong WF, Tyler-Smith C, Bainbridge M, Blackwell T, Zheng-Bradley X, Chen Y, Challis D, Clarke L, Ball EV, Cibulskis K, Cooper DN, Fulton B, Hartl C, Koboldt D, Muzny D, Smith R, Sougnez C, Stewart C, Ward A, Yu J, Xue Y, Altshuler D, Bustamante CD, Clark AG, Daly M, DePristo M, Flicek P, Gabriel S, Mardis E, Palotie A, Gibbs R. The functional spectrum of low-frequency coding variation. Genome Biol. 2011;12(9):R84. Epub 2011/09/16. doi: 10.1186/gb-2011-12-9-r84. PubMed PMID: 21917140; PMCID: PMC3308047.

24. 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;21(6):830-9. Epub 2011/04/05. doi: 10.1101/gr.115907.110. PubMed PMID: 21460062; PMCID: PMC3106316.

25. 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, Stutz 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. Mapping copy number variation by population-scale genome sequencing. Nature. 2011;470(7332):59-65. Epub 2011/02/05. doi: 10.1038/nature09708. PubMed PMID: 21293372; PMCID: PMC3077050.

26. 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;29(6):512-20. Epub 2011/05/10. doi: 10.1038/nbt.1852. PubMed PMID: 21552272; PMCID: PMC3270583.

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