# Amit R. Indap

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Specialties: Bioinformatics and Human Genetics

# Experience

Since July 2014 Genetics Research Scientist Good Start Genetics Cambridge, Massachusetts

- Performing bioinformatic and population genetic analyses on Ilumina sequencing data sets on tens of thousands of samples
- Evaluate and recommend software for improvements of bioinformatics pipeline using latest state-of-the-art methods
- Provide expertise in bioinformatic interpretation of reportable variants for clinical laboratory directors

December 2013 - July 2014 Postdoctoral Fellow Reproductive Endocrine Unit Massachusetts General Hospital Boston, Massachusetts

- Analyzed whole exome sequencing datasets to investigate the genetic basis of reproductive endocrine disorders
- Applied bioinformatic and statistical genomics methods to prioritize potential pathogenic alleles
- Collaborated with an interdisciplinary group of physicians, genetic counselors, and basic scientists

January 2009 - October 2013 PhD in Bioinformatics

Advisor Gabor T. Marth Boston College Chestnut Hill, Massachusetts

- Utilized existing and wrote novel software to analyze data from population scale and family based whole genome and whole exome resequencing studies to characterize the contribution of rare variants
- Analyzed capture sequencing data from the 1000 Genomes Pilot Project
- Identified a potential causal mutation in a Mendelian form of hearing loss
- Demonstrated the applicability of whole-genome amplified DNA in capture sequencing
- Implemented pedigree aware variant calling algorithm using graphical models

## August 2005 - December 2008 Bioinformatics Programmer

Cornell University Ithaca, New York

- Implemented bioinformatic data analysis pipelines for DNA re-sequencing and genotyping datasets for research projects in population and evolutionary genomics
- Contributed to the publication of over 10 papers in leading peerreviewed journals

#### February 2001 - July 2002 Bioinformatics Programmer

Stanford University Stanford, CA

• Implemented for sequencing and genotyping of candidate genes involved in complex diseases

## Skills

Bioinformatics: GATK, MOSAIK, Freebayes, PLINK, samtools, Picard

Programming: Python, R, Perl, familiar with Java and C++

Databases: MongoDB, MySQL

Molecular Biology: PCR, gel electrophoresis

# **Publications**

- [1] **Indap, A. R.**, Cole R., C. L. Runge, G. T. Marth, and M. Olivier. Variant discovery in targeted resequencing using whole genome amplified DNA. *BMC Genomics*, 14:468, 2013. [PubMed:23837845].
- [2] G. T. Marth, F. Yu, Indap, A. R., K. Garimella, S. Gravel, W. F. Leong, C. Tyler-Smith, M. Bainbridge, R. Gibbs, and the 1000 Genomes Consortium. The functional spectrum of low-frequency coding variation. *Genome Biol.*, 12(9):R84, 2011. [PubMed Central:PMC3308047] [DOI:10.1186/gb-2011-12-9-r84] [PubMed:21917140].
- [3] S. Gravel, B. M. Henn, R. N. Gutenkunst, Indap, A. R., G. T. Marth, A. G. Clark, F. Yu, R. A. Gibbs, C. D. Bustamante, and the 1000 Genomes Consortium. Demographic history and rare allele sharing among human populations. *Proc. Natl. Acad. Sci. U.S.A.*, 108(29):11983–11988, Jul 2011. [PubMed Central:PMC3142009] [DOI:10.1073/pnas.1019276108] [PubMed:21730125].
- [4] 1000 Genomes Consortium. A map of human genome variation from population-scale sequencing. Nature, 467(7319):1061–1073, Oct 2010. [PubMed Central:PMC3042601] [DOI:10.1038/nature09534] [PubMed:20981092].
- [5] A. M. Andres, M. J. Hubisz, Indap, A., D. G. Torgerson, J. D. Degenhardt, A. R. Boyko, R. N. Gutenkunst, T. J. White, E. D. Green, C. D. Bustamante, A. G. Clark, and R. Nielsen. Targets of balancing selection in the human genome. *Mol. Biol. Evol.*, 26(12):2755–2764, Dec 2009. [PubMed Central:PMC2782326] [DOI:10.1093/molbev/msp190] [PubMed:19713326].
- [6] D. G. Torgerson, A. R. Boyko, R. D. Hernandez, Indap, A., X. Hu, T. J. White, J. J. Sninsky, M. Cargill, M. D. Adams, C. D. Bustamante, and A. G. Clark. Evolutionary processes acting on candidate cis-regulatory regions in humans inferred from patterns of polymorphism and divergence. *PLoS Genet.*, 5(8):e1000592, Aug 2009. [PubMed Central:PMC2714078] [DOI:10.1371/journal.pgen.1000592] [PubMed:19662163].
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- [8] A. Auton, K. Bryc, A. R. Boyko, K. E. Lohmueller, J. Novembre, A. Reynolds, Indap, A., M. H. Wright, J. D. Degenhardt, R. N. Gutenkunst,

- K. S. King, M. R. Nelson, and C. D. Bustamante. Global distribution of genomic diversity underscores rich complex history of continental human populations. *Genome Res.*, 19(5):795–803, May 2009. [PubMed Central:PMC2675968] [DOI:10.1101/gr.088898.108] [PubMed:19218534].
- [9] M. R. Nelson, K. Bryc, K. S. King, Indap, A., A. R. Boyko, J. Novembre, L. P. Briley, Y. Maruyama, D. M. Waterworth, G. Waeber, P. Vollenweider, J. R. Oksenberg, S. L. Hauser, H. A. Stirnadel, J. S. Kooner, J. C. Chambers, B. Jones, V. Mooser, C. D. Bustamante, A. D. Roses, D. K. Burns, M. G. Ehm, and E. H. Lai. The Population Reference Sample, POPRES: a resource for population, disease, and pharmacological genetics research. Am. J. Hum. Genet., 83(3):347–358, Sep 2008. [PubMed Central:PMC2556436] [DOI:10.1016/j.ajhg.2008.08.005] [PubMed:18760391].
- [10] J. Novembre, T. Johnson, K. Bryc, Z. Kutalik, A. R. Boyko, A. Auton, Indap, A., K. S. King, S. Bergmann, M. R. Nelson, M. Stephens, and C. D. Bustamante. Genes mirror geography within Europe. *Nature*, 456(7218):98–101, Nov 2008. [PubMed Central:PMC2735096] [DOI:10.1038/nature07331] [PubMed:18758442].
- [11] R. Blekhman, O. Man, L. Herrmann, A. R. Boyko, Indap, A., C. Kosiol, C. D. Bustamante, K. M. Teshima, and M. Przeworski. Natural selection on genes that underlie human disease susceptibility. *Curr. Biol.*, 18(12):883–889, Jun 2008. [PubMed Central:PMC2474766] [DOI:10.1016/j.cub.2008.04.074] [PubMed:18571414].
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- [13] K. E. Lohmueller, Indap, A. R., S. Schmidt, A. R. Boyko, R. D. Hernandez, M. J. Hubisz, J. J. Sninsky, T. J. White, S. R. Sunyaev, R. Nielsen, A. G. Clark, and C. D. Bustamante. Proportionally more deleterious genetic variation in European than in African populations. *Nature*, 451(7181):994–997, Feb 2008. [PubMed Central:PMC2923434] [DOI:10.1038/nature06611] [PubMed:18288194].
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- [15] Rhesus Macaque Genome Consortium. Evolutionary and biomedical insights from the rhesus macaque genome. *Science*, 316(5822):222–234, Apr 2007. [DOI:10.1126/science.1139247] [PubMed:17431167].
- [16] Indap, A. R., G. T. Marth, C. A. Struble, P. Tonellato, and M. Olivier. Analysis of concordance of different haplotype block partitioning algorithms. *BMC Bioinformatics*, 6:303, 2005. [PubMed Central:PMC1343594] [DOI:10.1186/1471-2105-6-303] [PubMed:16356172].

## Education

**2009-2013** *PhD in Bioinformatics at Boston College* Chestnut Hill, Massachusetts

Title: Discovering rare variants from populations to families. Advisor: Gabor T. Marth

**2003-2005** MS in Bioinformatics at Medical College of Wisconsin and Marquette University Milwaukee, Wisconsin

Title: Analysis of concordance of haplotype block partitioning algorithms. Advisor: Michael Olivier

1996-2000 BS in Molecular and Cellular Biology, minor Computer Science University of Arizona Tucson, Arizona

Selected as Outstanding Graduating Senior in Department of Molecular and Cellular Biology