# Amit R. Indap

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Specialties: Bioinformatics, Human Genetics, Python

## Experience

Since December 2013 Research Fellow for Reproductive Endocrine Unit, Massachusetts General Hospital Boston, Massachusetts

Investigating the genetic basis of reproductive endocrine disorders by whole exome and genome sequencing. Applying bioinformatic and statistical genomics methods to prioritize potential causal alleles. Working in an interdisciplinary group of physicians, genetic counselors, and basic scientists.

January 2009 - October 2013 PhD in Bioinformatics with Gabor T. Marth Boston College Chestnut Hill, Massachusetts

Utilized existing and wrote novel software to analye data from population scale and family based whole genome and whole exome re-sequencing studies to characterize the contribution of rare variants to human genetic diversity and disease risk. Projects included analyzing capture sequencing data from the 1000 Genomes Pilot Project, identifying a potential causal mutation in a Mendelian disease, demonstrating the applicability of whole-genome amplified DNA in capture sequencing, and implementing a pedigree aware variant calling algorithm.

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 which helped facilitate the publication of over 10 papers in leading scientific journals.

February 2001 - July 2002 Bioinformatics Programmer at Stanford University Stanford, CA

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

### **Selected Publications**

Variant discovery in targeted resequencing using whole genome amplified DNA The functional spectrum of low-frequency coding variation

A map of human genome variation from population-scale sequencing

Assessing the evolutionary impact of amino acid mutations in the human genome

Proportionally more deleterious genetic variation in European than in African populations

Analysis of concordance of different haplotype block partitioning algorithms

#### Skills

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

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

Molecular Biology: PCR, gel electrophoresis

### Education

 ${\bf 2009\text{-}2013}\ PhD$  in Bioinformatics at  $Boston\ College$  Chestnut Hill , Massachusetts

Title: Discovering rare variants from populations to families. Advisor: Gabor T. Marth Defended in October 2013

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