Amit R. Indap

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Specialties: Bioinformatics, Human Genetics, Variant Curation and Reporting

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Experience

Research Scientist

Human Longevity Inc. San Diego, California September 2015 - current

- Implementing variant reporting pipelines for pharmacogenetics and autoimmune diseases for HLI whole genome and exome reports using Docker and AWS
- Investigating distribution and frequency of loss-of-function variants in HLI cohort of samples

Genetics Research Scientist

Good Start Genetics Cambridge, MA July 2014 - August 2015

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

Research Fellow

Massachusetts General Hospital Boston, MA December 2013 - July 2014

- 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
- Worked in an interdisciplinary group of physicians, genetic counselors, and basic scientists

Graduate Research Assistant

Boston College Chestnut Hill, MA January 2009 - October 2013

- Utilized existing and wrote novel software to analyze data from population scale and family based whole genome and whole exome re-sequencing 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

Bioinformatics Programmer

Cornell University Ithaca, NY August 2005 - December 2008

- 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 peer-reviewed journals

Bioinformatics Programmer

Stanford University Stanford, CA February 2001 - July 2002

- Implemented 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

Full list of publications listed here

Skills

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

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

Databases : MongoDB, MySQL

IT/Computer : AWS EC2, S3, SWS, Docker, git

Molecular Biology : PCR, gel electrophoresis

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