

# Asif Javed

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## Research Interests

Computational Genomics, Translational and Clinical Genetics, Next Generation Sequencing

## Work Experience

**Research Scientist** August 2014 - present  
Genome Institute of Singapore, A\*STAR Singapore

**Research Associate** December 2011 - July 2014  
Genome Institute of Singapore, A\*STAR Singapore

*Design and application of analysis methods which combine clinical reports with sequencing data to predict disease causal variant(s) in rare disorders (<http://phen-gen.org/>)*

Published in **Nature Methods**, first and co-corresponding author.

- Investigator for **2M SGD BMRC** grant for industrial collaboration with **Appistry Inc.** (USA) to study and compare NGS pipelines
- Principal investigator for **160k SGD** research collaborative agreement with **Ramathibodi hospital**, (Mahidol University, Thailand) analyzing rare familial disorders
- Principal investigator for GIS team participating international genomic interpretation competition **CLARITY** challenge (**Genome Biology**)
- Conducted multiple 2-day Next Generation Sequencing Bioinformatics workshops  
UITM Malaysia (Apr 12), GIS A\*STAR (May 12), AIT Biotech (Nov 12)
- Funded and supervised one research officer (2 years), trained 4 interns

**Postdoctoral Researcher** December 2008 - December 2011  
Computational Biology Center, IBM T. J. Watson Research Yorktown, NY, USA

*Designed computational tool to identify historic recombinations from their evidence in SNP data from current individuals. Frequency and sharing of recombination elucidate ancient human migration paths*

- Received **IBM Research Accomplishment** for *Results from Genographic*
- Received **IBM First Invention Filing Award**, for US patent *Sampling the space of Ancestral Recombination Graphs*

**Research Intern** May 2008 - August 2008  
Computational Biology Center, IBM T. J. Watson Research Yorktown, NY, USA

**Teaching / Research Assistant** August 2004 - December 2008  
Rensselaer Polytechnic Institute Troy, NY, USA

*Designed linear algebraic algorithms to select a small panel of representative markers (tagging SNPs) which can be used to predict the remaining from SNP-chip data*

*Courses assisted:* Computer Science 1, Models of Computation, Randomized Algorithms

**Teaching / Research Assistant** August 2001 - May 2004  
University of Illinois at Chicago Chicago, IL, USA

*Designed and implemented distributed data mining algorithms*

*Courses assisted:* Random Signal Processing, Analog Communication Circuits, Computer Architecture, Digital Logic

### Teaching Assistant

August 2000 - August 2001

Lahore University of Management and Sciences

Lahore, Pakistan

*Courses assisted:* Discrete Mathematics, Algorithm Design, Advanced Algorithm Design, Circuits and Systems 1, Algorithms in Computational Biology

### Clinical Collaborations

Studying the following disorders in clinical collaborations within and outside Singapore

- Leukemia progression
- Intellectual disability
- Novel Ophthalmologic condition
- Muscular dystrophies
- Cardiac disorders
- Neuro-developmental disorders

### Education

**Rensselaer Polytechnic Institute**

Troy, NY, USA

Ph.D., Computer Science, (CGPA 4.0), December 2008

thesis: *A computational analysis of human genetic variation*

**University of Illinois at Chicago**

Chicago, IL, USA

M.S., Computer Engg., (CGPA 3.9, C.S. GPA 4.0), May 2004

thesis: *Scalable parallel algorithm for mining frequent patterns on message passing systems*

**University of Engineering and Technology, Lahore**

Lahore, Pakistan

B.S. in Electrical Engineering (with honors) 2000

### Publications

- B. Pupaçdi, **A. Javed**, M.J. Zaki and M. Ruchirawat, *NSIT: Novel Sequence Identification Tool*, **PLoS One**, Sep 2014. (**IF=3.53**)
- **A. Javed**<sup>\*</sup>, S. Agrawal, and P.C. Ng<sup>\*</sup>, *Phen-Gen: combining Phenotype and Genotype to analyze rare disorders*, **Nature Methods**, Sep 2014. (<sup>\*</sup> corresponding authors) (**IF=23.56**)
- C.A. Brownstein, ..., **A. Javed**, et al., *An international effort towards developing standards for best practices in analysis, interpretation and reporting of clinical genome sequencing results in the CLARITY Challenge*, **Genome Biology**, Mar 2014. (**IF=10.50**)
- **A. Javed**, M. Mele, M. Pybus, et. al, and The Genographic Consortium, *Recombination networks as genetic markers: a human variation study of the Old World*, **Hum Genet**, Apr 2012. (**IF=4.63**)
- M. Mele, **A. Javed**, M. Pybus, et. al, and The Genographic Consortium, *The footprint of recombination gives a new insight in the effective population size and the history of the Old World human populations*, **Mol Biol Evol**, Jan 2012. (**IF=14.31**)
- **A. Javed**, P. Paschou, M.W. Mahoney, and P. Drineas, *Efficient genomewide selection of PCA-correlated tSNPs for genotype imputation.*, **Ann Hum Genet**, Nov 2011. (**IF=1.93**)
- **A. Javed**, M. Pybus, M. Mele, et. al, *IRiS: Construction of ARG networks at genomic scales*, **Bioinformatics**, Sep 2011. (**IF=5.32**)
- L. Parida, P.F. Palamara, **A. Javed**, *A Minimal Descriptor of an Ancestral Recombinations Graph*, **BMC Bioinformatics**, APBC, Feb 2011. (**IF=2.67**)

- P. Paschou, J. Lewis, **A. Javed**, P. Drineas, *Ancestry informative markers for fine-scale individual assignment to worldwide populations*, **J Med Genet.** Dec 2010. (IF=5.64)
- M. Mele, **A. Javed**, F. Calafell, et. al, and The Genographic Consortium, *A New Method to Reconstruct Recombination Events at a Genomic Scale*, **PLOS Comput Biol.** Nov 2010. (IF=4.87)
- **A. Javed** and L. Parida, *Recombinomics: Population Genomics from a Recombination Perspective*, **Proceedings of C3S2E**, May, 2010. (Acceptance=23%)
- L. Parida, **A. Javed**, M. Mele, F. Calafell, J. Bertranpetit and The Genographic Consortium, *Minimizing recombinations in consensus networks for phylogeographic studies*, **BMC Bioinformatics**, Jan 2009. (IF=2.67)
- L. Parida, **A. Javed**, M. Mele, and J. Bertranpetit, *A case for Recombinomics*, IBM Technical Report RC24677, Aug 2008.
- **A. Javed** and P. Paschou, *Extracting tagging SNPs from genome-wide datasets*, DMBI, workshop held in conjunction with SDM, Apr 2007.
- P. Paschou, M.W. Mahoney, **A. Javed**, et. al, *Intra- and inter-population genotype reconstruction from tagging SNPs*, **Genome Research**, Jan 2007. (IF=14.4)
- P. Drineas, **A. Javed**, M. Magdon-Ismael, G. Pandurangan, R.Virrankoski, and A. Savvides, *Distance matrix reconstruction from incomplete distance information for sensor network localization*, (SECON), Sep 2006. (Acceptance=26%)
- **A. Javed** and A. Khokhar, *Frequent pattern mining on message passing multiprocessor systems*, (Distributed and Parallel Databases), Nov 2004. (IF=0.81)
- **A. Javed** and A. Khokhar, *Scalable parallel algorithm for mining frequent patterns on message passing parallel systems*, ISCA (PDCS), Aug 2003.

### **Consortium publications**

- G. Brandt et al., and **The Genographic Consortium\***, *Ancient DNA reveals key stages in the formation of central European mitochondrial genetic diversity*, **Science**, Oct 13. (IF=31.48)
- P. Brotherton et al., and **The Genographic Consortium\***, *Neolithic mitochondrial haplogroup H genomes and the genetic origins of Europeans*, **Nat Commun**, Apr 2013. (IF=10.02)
- D.A. Badro et al., and **The Genographic Consortium\***, *Y-chromosome and mtDNA genetics reveal significant contrasts in affinities of modern Middle Eastern populations with European and African populations*, **PLoS One**, Jan 2013. (IF=3.53)
- G. Arunkumar et al., and **The Genographic Consortium\***, *Population differentiation of southern Indian male lineages correlates with agricultural expansions predating the caste system*, **PLoS One**, Nov 2012. (IF=3.53)
- B. Martnez-Cruz et al., and **The Genographic Consortium\***, *Evidence of pre-roman tribal genetic structure in basques from uniparentally inherited markers*, **Mol Biol Evol**, Sep 2012. (IF=14.31)
- B. Martnez-Cruz et al., and **The Genographic Consortium\***, *Y-chromosome analysis in individuals bearing the Basarab name of the first dynasty of Wallachian kings*, **PLoS One**, Jul 2012. (IF=3.53)

- T.G. Schurr et al., and **The Genographic Consortium\***, *Clan, language, and migration history has shaped genetic diversity in Haida and Tlingit populations from Southeast Alaska*, **Am J Phys Anthropol**, Jul 2012. (IF=2.51)
- M.C. Dulik et al., and **The Genographic Consortium\***, *Y-chromosome analysis reveals genetic divergence and new founding native lineages in Athapaskan- and Eskimoan-speaking populations*, **Proc Natl Acad Sci**, May 2012. (IF=9.81)
- M. Haber et al., and **The Genographic Consortium\***, *Afghanistan's ethnic groups share a Y-chromosomal heritage structured by historical events*, **PLoS One**, Mar 2012. (IF=3.53)
- Y. Lu et al., and **The Genographic Consortium\***, *Mitochondrial origin of the matrilineal Mosuo people in China*, **Mitochondrial DNA**, Feb 2012. (IF=1.27)
- L. Kang et al., and **The Genographic Consortium**, *Y-chromosome O3 haplogroup diversity in Sino-Tibetan populations reveals two migration routes into the eastern Himalayas*, **Ann Hum Genet**, Jan 2012. (IF=1.93)
- J.B. Gaieski et al., and **The Genographic Consortium\***, *Genetic ancestry and indigenous heritage in a Native American descendant community in Bermuda*, **Am J Phys Anthropol**, Nov 2011. (IF=2.51)
- M.S. Jota et al., and **The Genographic Consortium\***, *A new subhaplogroup of native American Y-Chromosomes from the Andes*, **Am J Phys Anthropol**, Sep 2011. (IF=2.51)
- S. Yan et al., and **The Genographic Consortium\***, *An updated tree of Y-chromosome Haplogroup O and revised phylogenetic positions of mutations P164 and PK4*, **Eur J Hum Genet**, Sep 2011. (IF=4.32)
- X. Cai et al., and **The Genographic Consortium\***, *Human Migration through Bottlenecks from Southeast Asia into East Asia during Last Glacial Maximum Revealed by Y Chromosomes*, **PLoS One**, Aug 2011. (IF=3.53)
- B. Martnez-Cruz et al., and **The Genographic Consortium\***, *Multiplex single-nucleotide polymorphism typing of the human Y chromosome using TaqMan probes*, **Investig Genet**, May 2011. (IF=5.8)
- O. Balanovsky et al., and **The Genographic Consortium\***, *Parallel Evolution of Genes and Languages in the Caucasus Region*, **Mol Biol Evol**, May 2011. (IF=14.31)
- C.J. Adler et al., and **The Genographic Consortium\***, *Survival and recovery of DNA from ancient teeth and bones*, **Journal of Archaeological Science**, May 2011. (IF=2.14)
- K. Yang et al., and **The Genographic Consortium\***, *Positive selection on mitochondrial M7 lineages among the Gelong people in Hainan*, **Journal of Human Genetics** March 2011. (IF=2.36)
- M. Haber et al., and **The Genographic Consortium\***, *Influences of history, geography, and religion on genetic structure: the Maronites in Lebanon*, **Eur J Hum Genet** Dec 2010. (IF=4.32)
- Z. Qin et al., and **The Genographic Consortium\***, *A mitochondrial revelation of early human migrations to the Tibetan Plateau before and after the last glacial maximum*, **Am J Phys Anthropol** Dec 2010. (IF=2.51)
- W. Haak et al., and **The Genographic Consortium\***, *Ancient DNA from European early neolithic farmers reveals their near eastern affinities*, **PLOS Biol.** Nov 2010. (IF=12.69)

## Patents

- *Sampling the space of Ancestral Recombination Graphs* (US 20120331008 A1).

## Selected Presentations

- *Incorporating phenotypic information to improve Mendelian disease-gene predictions*, poster presented at **63rd Annual Meeting of ASHG**, Oct 2013.
- *Leveraging phenotypic priors for disease-gene implication in rare disorders*, poster presented at HGM ICG joint conference April 2013.
- *Two roads both taken: walking the phenotypic and genotypic paths to disease gene implication*, poster presented at the **62th Annual Meeting of ASHG**, Nov. 2012.
- *Leveraging phenotypic priors for disease-gene implication in rare disorders*, invited talk at **National University Health System**, Singapore Oct. 2012.
- *Recombinomics*, oral presentation at Structure Discovery in Biology: Motifs, Networks & Phylogenies seminar at **Schloss Dagstuhl**, Jun. 2010.
- *Recombinomics*, invited talk at **Genome Biology and Bioinformatics Annual Retreat** at University of Toronto, May 2010.
- *RECO project*, oral presentation at Genographic Consortium annual meeting, Feb. 2010.
- **A. Javed**, *Recombinomics: recombination-based population genomics*, accepted tutorial at **APBC 2010**, Jan. 2010.
- *IRiS-detecting recombinations for phylogenetics and more*, oral presentation at Genome Informatics meeting at **CSHL**, Oct. 2009.
- *Recombinations-based population genomics*, oral presentation at Workshop on Algorithms in Human Population Genomics at **DIMACS**, Rutgers University, Apr. 2009.
- *Novel matrix decomposition for knowledge discovery in genomic data*, white paper presentation at **Cyber-Enabled Discovery and Innovation**, NSF symposium held at Rensselaer Polytechnic Institute, Sep. 2007.
- *Parallel frequent pattern mining*, poster presentation in **Midwest Database Symposium**, Chicago April 2004.

## Computer Skills

*Operating Systems:* Microsoft Windows, Unix, Linux

*Programming Languages:* C/C++, Assembly (x86 architecture), Perl

*Parallel Programming:* MPI 1 & 2, OpenMP

*Tools:* Matlab, bash shell scripting, L<sup>A</sup>T<sub>E</sub>X

*Bioinformatics:* BWA, Bowtie, SAMtools, GATK, IGV, Picard, VCFtools, bedtools, ANNOVAR, VAAST, PLINK, arlequin, Beagle, LDHat, fastPHASE, IMPUTE2

## Professional Activities

- Program Committee Member, RECOMB-CG 14, ISMB 13, BICoB 13, AIMM 12, BIOKDD (KDD 12), BICoB 12, BIOKDD (KDD 11), BIODM (ICDM 10), DMBI (SDM 08)
- Reviewer for Bioinformatics, Human Genetics, PloS Genetics, Nature Methods.

## References

Available on request.