

Karan Ajay Kapuria

510-309-0140

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PROFILE

Bioinformatics professional with 3 years of experience in next-generation sequencing (NGS) pipeline development, validation, and implementation.

EDUCATION

- Georgia Institute of Technology**, Atlanta, GA 2016 – 2017
Master of Science, Bioinformatics
- Guru Nanak Khalsa College**, Mumbai, India 2014 – 2016
Master of Science, Bioinformatics
- Mithibai College**, Mumbai, India 2011 – 2014
Bachelor of Science, Chemistry

PROFESSIONAL EXPERIENCE

Laboratory Corporation of America (*MNG Labs became part of LabCorp*)

Atlanta, GA

Biostatistician

March 2019 – Present

- Evaluate analytical performance, maintain and develop new modules for all MNG's NGS analysis workflows (WES/WGS/Targeted Panels/Mitochondrial DNA)
- Provide bioinformatics support to reporting specialists with case specific queries
- Work closely with Director of R&D and Director of Reporting to update and improve bioinformatics reporting functionalities
- Automate MNG's flagship NGS pipeline with the goal of combining multiple-cloud technologies to reduce workflow debts and yield a flexible platform in link with other databases.
- Work directly with the LabCorp's NGS team to present MNG's bioinformatics capabilities to LabCorp, addressing their needs.

MNG Laboratories

Atlanta, GA

Bioinformatics Developer

March 2018 – March 2019

- Provided first-in-class bioinformatics services for a CAP/CLIA accredited clinical NGS lab specializing in neurogenetics utilizing Illumina technologies (MiSeq, HiSeq 1500/2500, NovaSeq 6000, BaseSpace Sequencing Hub)
- Lead bioinformatics development and provide support on bringing new NGS products (WGS/Express Exome) to market, working closely with Director of Bioinformatics, CMO, and CIO
- Develop / validate / and lead implementation of NGS pipelines on Amazon Web Services (AWS), including:
 - Whole genome sequencing: constitutional whole genome variant, copy number, expansion repeat, uniparental disomy, Mitochondrial heteroplasmy / deletion / depletion analysis
 - Whole transcriptome sequencing: differential expression, junction, variant analysis
 - Whole exome sequencing: constitutional exome variant, copy number, uniparental disomy analysis
 - Custom genetics panel: constitutional variant, single exon resolution copy number analysis
 - Mitochondrial analysis: heteroplasmy (1%) variant analysis, break point resolution deletion analysis with quantification

Georgia Institute of Technology (Dr. Brown's Lab)

Atlanta, GA

Bioinformatics Data Analyst

April 2017 – December 2017

Graduate Research Assistant

August 2016 – April 2017

- Developed and managed a metagenomics OTU classification pipeline using QIIME and DADA2 for 16s rRNA sequencing data.
- Performed PCA, machine learning and regression to evaluate co-occurrence patterns of in microbiome abundance data.
- Creating visualizations using R package, ggplot2 and phyloseq. fitting the dataset to generalized lotka-volterra model.
- Performed network analysis in lung microbiome using SparCC and Pearson co-relations.
- Performed machine learning on cross sectional abundance matrix obtained from lung microbiome in Cystic Fibrosis (CF) affected cohort.

Guru Nanak Institute of Research and Development

Mumbai, India

Research Assistant

June 2014 - March 2016

- Designed anti-psoriatic flavonoid analogues using Chem Sketch and docked against protein target using GOLD (Genetic Optimization for Ligand Docking) software.
 - Performed and lead the QSAR study on best binding ligands obtained from GOLD using VLifeMDS software for predicting models with best biological activities.
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SKILLS

Technology

- Software Languages: Bash, R Statistical Programming Language, Python, SQL, C++
- Software Tools: Illumina Base Space Sequencing Hub (BSSH), Genome Analysis Toolkit (GATK), Dragen, NextGENe, Burrows-Wheeler Aligner (BWA), Integrative Genomics Viewer (IGV), VarScan, Picard, bedtools, vcftools, bcftools, samtools, Annovar, Sun Grid Engine, Variant Effect Predictor (VEP)
- Amazon Web Services (AWS) : EC2, S3, ECR, AWS Batch, ECS, SNS, IAM, CLI, boto3 (python for AWS)
- Online Resources / Databases: GTEx, Online Mendelian Inheritance in Man (OMIM), ClinVar, Catalogue Of Somatic Mutations In Cancer (COSMIC), Ensembl, 1000 Genomes, genome Aggregation Database (gnomAD), Exome Aggregation Consortium (ExAC), dbSNP, dbVAR, MITOMAP, STRING (Protein-Protein Interaction Networks), International Mouse Phenotyping Consortium (IMPC), SIFT, PolyPhen, UCSC Genome Browser
- Management Tools and Version Control: Jira, Bitbucket, Github

SCHOLARSHIPS and AWARDS

- Computational Biology Faculty Research Awards – Georgia Institute of Technology
For research proposal on ‘**Network Analysis of Microbial Communities in Cystic Fibrosis Lung**’. *Fall 2017*
- Computational Biology Faculty Research Awards – Georgia Institute of Technology
For research proposal on ‘**Inferring Interactions in the Cystic Fibrosis Microbiome with Dynamic Modeling and Machine Learning**’. *Spring 2017*

PUBLICATIONS and POSTERS

Co-author. **Determination of the phylogenetic origins of the Árpád Dynasty based on Y chromosome sequencing of Béla the Third**. <https://doi.org/10.1038/s41431-020-0683-z>, *Nature*, 07 July 2020

Co-presented poster. **The Role of Whole Genome Sequencing in Clinical Diagnosis**. 2018. The American Society of Human Genetics.

Co-presented poster. **Complementary Test Methods Needed to Improve Clinical Sensitivity of Genetic Testing**. 2018. The American Society of Human Genetics.

Co-presented poster. **Applying Machine Learning Techniques on Cystic Fibrosis Lung Microbiome**. 2017. Metagenomics Symposium, Georgia Institute of Technology.