Pooja Pravinbabu

Genome/ Bioinformatics Analyst

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PROFILE

As a genome analyst, I specialize in variant interpretation following ACMG guidelines, utilizing my skills in bioinformatics, and programming in R and Python to manage and analyze large genomic datasets. My expertise is crucial in diagnosing genetic disorders. Dedicated to continuous learning, I aim to stay at the forefront of technological advancements in genomics, contributing significantly to the field and improving patient outcomes through precise and accurate genomic analysis.

PUBLICATIONS

Pravinbabu, P., Holla, V.V., Phulpagar, P. et al. A splice altering variant in NDRG1 gene causes Charcot-Marie-Tooth disease, type 4D. Neurol Sci (2022)
SpringerLink
11/02/2022

PROFESSIONAL EXPERIENCE

Sandor Bioinformatics Private Limited
09/2023 – present | BANGALORE, India
Bioinformatics Analyst

- Proficient in NGS data analysis and variant calling, using tools like BWA and GATK.
- Expertise in variant identification, annotation, and prioritization based on functional impact and disease relevance.
- Skilled in utilizing genomics databases (NCBI, Ensembl, UCSC, CLINVAR, LITVAR) and bioinformatics tools (Franklin, Mutalyzer, Splice AI) for comprehensive genomic analysis.

Medgenome
01/2022 – 09/2023 | BANGALORE, India
Genome Analyst

- **Variant Interpretation**: Expertise in identifying and annotating genetic variants, including SNPs, INDELs, and structural variants. Proficient in variant prioritization based on functional impact, population frequency, and disease relevance. Proficient in applying ACMG criteria to classify variants based on pathogenicity, incorporating factors such as population frequency, functional impact, and clinical significance. Track record of accurately classifying variants, contributing to accurate genetic diagnosis and clinical decision-making.
- **Bioinformatics**: Proficient in using databases like NCBI, Ensembl, UCSC, CLINVAR, LITVAR. And proficient in using tools like Franklin, Mutalyzer, Splice AI etc.

Institute of Bioinformatics
11/2020 – 01/2022 | BANGALORE, India
Research Trainee under Dr.Babylakshmi Muthuswamy

- Proficient in NGS data analysis, variant calling (using tools like BWA, GATK), and bioinformatics tasks including data preprocessing, and alignment with databases like NCBI, Ensembl.
- Expertise in identifying and prioritizing genetic variants, with knowledge in statistical methods, GWAS, and pathway analysis tools.
- Skilled in programming (Python, R), version control (Git), and data visualization for informative plots and figures.
- Additionally, experienced in proteomic data analysis, biomarker discovery, and protein homology modeling.

EDUCATION

M.Sc Genetics
Center for Human Genetics
06/2018 – 08/2020 |
BANGALORE, India
Dissertation Project: Allelic Heterogeneity in COL7A1 in Epidermolysis Bullosa Condition
Supervisor: Dr. Ravi Hiramagalor

B.Sc Biology with Honours, Minor in Data Science
Azim Premji University
06/2015 – 05/2018 | BANGALORE, India
Honours Thesis: Memory Transmission Following Amputation in Schmidtea Mediterranea
Supervisor: Dr. Sravanti Uppaluri

AWARDS

APOGEE fest
Birla Institute of Technology
Participated in a paper presentation contest. Paper written as a part of B.Sc honors project (Memory Transmission Following Amputation in Schmidtea Mediterranea) got selected for APOGEE fest and got 1st price in paper presentation event (BITS Pilani)

CERTIFICATES

- SAS BASE PROGRAMMING SPECIALIST
- Certificate of completion in Full Stack Development

SKILLS

Genetic Knowledge ●●●●○
Profound understanding of genetics, molecular biology, and genomic sciences. Familiarity with genetic disorders, inheritance patterns, and gene function.

Bioinformatics ●●●●○
Ability to use bioinformatics tools and databases effectively. This includes software for DNA sequencing analysis, gene prediction, functional annotation, and comparative genomics.

ACMG Guidelines ●●●●○
Knowledge of standards and protocols for interpreting genetic variants, such as those provided by the American College of Medical Genetics and Genomics (ACMG), is crucial for clinical application.

Programming Skills ●●●○○
Fluency in programming languages like Python and R. These skills are necessary for writing scripts to automate tasks, manage data, and perform complex analyses.

Communication Skills ●●●●○
Strong ability to communicate complex genetic information clearly and effectively to a variety of audiences, including non-specialists, clinicians, and researchers.

Attention to Detail ●●●●●
Genome analysis requires a meticulous approach to ensure accuracy and reliability in data interpretation and reporting.