UNIVERSITY OF MYSORE

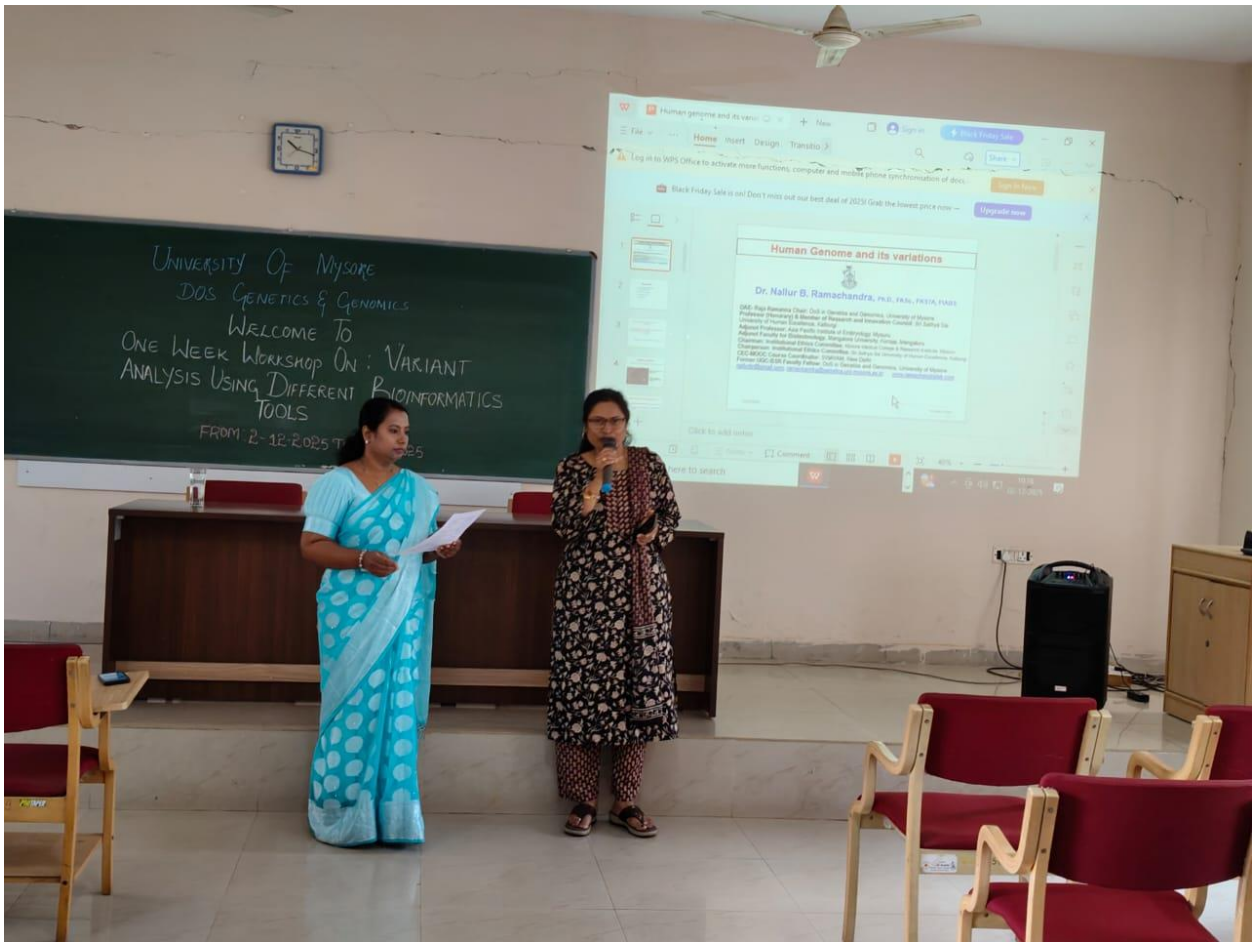
Department Name: Department of Studies in Genetics and Genomics

MANASAGANGOTRI, MYSORE-570 006

RUSA SPONSORED One Week WORKSHOP: Variant Analysis Using Different Bioinformatics Tools

From 02-12-2025 to 08-12-2025

The one week workshop on Hands on Training on "**Variant Analysis Using Different Bioinformatics Tools"** RUSA sponsored Faculty improvement program in association with Department of Genetics and Genomics, University of Mysore. Held from 02nd to 08th December 2025 at the Department of Genetics and Genomics, the workshop was attended by a diverse group of participants,, faculty members and invitees. This workshop was officially inaugurated and welcomed the participants by Prof. Suttur S Malini, chairperson, Department of Studies in Genetics and Genomics .This workshop was a great platform for knowledge sharing among the participants and attendees.



Session: 1
Topic: Human Genome and It's Variants
Resource Person: Prof. N.B. Ramachandra
Designation: DAE Rajaramanna Chair, DoS in Genetics and Genomics, UOM
Time: 10.00Am to 11.30AM

The first session of the one-week workshop titled "Variant Analysis Using Different Bioinformatics Tools", held at the Department of Studies in Genetics and Genomics, University of Mysore, began with a detailed lecture by Prof. Nallur B. Ramachandra on "Human Genome and Its Variants." His talk introduced participants to the structure, complexity, and diversity of the human genome, while highlighting why genomic-level understanding is essential in modern biology and

medicine. Prof. Ramachandra described the human genome as a vast map containing our evolutionary history, genetic identity, and susceptibility to disease. Over the years, coordinated global projectsfrom the Human Genome Project to pangenome initiativeshave transformed how scientists read and interpret this map. The lecture provided the scientific foundation needed for the rest of the workshop by connecting genome science with variant analysis, population diversity, and clinical applications.

Key Points

Prof. Ramachandra began by explaining the genome as the complete DNA blueprint of an organism. To understand this blueprint, scientists first needed genetic mapslinkage maps, physical maps, and gene mapswhich tell us where genes lie, how chromosomes are organized, and how traits are inherited. This mapping effort laid the groundwork for the Human Genome Project (HssGP), launched in 1990. The HGP, completed in 2003, produced the first draft and later the nearcomplete sequence of human DNA. It revealed the presence of 23 chromosome pairs, identified genes like IGF2 (one of the smallest) and dystrophin (one of the largest), and showed the genome's immense complexity. This reference map became the global standard against which all human genetic variation is compared.

With a reference genome available, the next major question was: How do humans differ from one another? This led to the study of genetic variants.

The lecture focused on major variant types:

- Single Nucleotide Polymorphisms (SNPs), the most common variants used in population studies
- Indels, small insertions and deletions

- Microsatellites and trinucleotide repeats, associated with disorders like Huntington's
- Copy Number Variations (CNVs), large changes in DNA dosage
- SNPs received special emphasis because they form the basis of genome-wide association studies (GWAS) and provide insights into traits, disease risk, drug response, and population migration. CNVs, although fewer, have strong biological impact and help in studying conditions such as Parkinson's disease, autism, dyslexia, and asthma.

Session: 2
Topic: Next Generation Sequencing
Resource Person: Dr. Chaithra PT
Designation: Guest Faculty, DoS in Genetics and Genomics, UOM
Time: 11.45Am to 1.15PM

Introduction: A workshop titled "Variant Analysis Using Different Bioinformatics Tools" was conducted to equipparticipants with skills in genetic variant identification and computational analysis. As part of this workshop, Dr. Chaithra P. T. delivered a specialized session on Next Generation Sequencing. The session aimed to provide students and researchers with a comprehensive understanding of sequencing technologies, their evolution, and modern applications in biomedical research.

Objectives of the Session:

1. To introduce participants to the concept and evolution of NGS.
2. To explain how sequencing data is generated before variant analysis begins.
3. To describe NGS workflow and its importance in detecting genomic variants.
4. To connect sequencing technologies with downstream bioinformatics tools.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